

# A Trio of Visualization Design Studies

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**University of British Columbia**

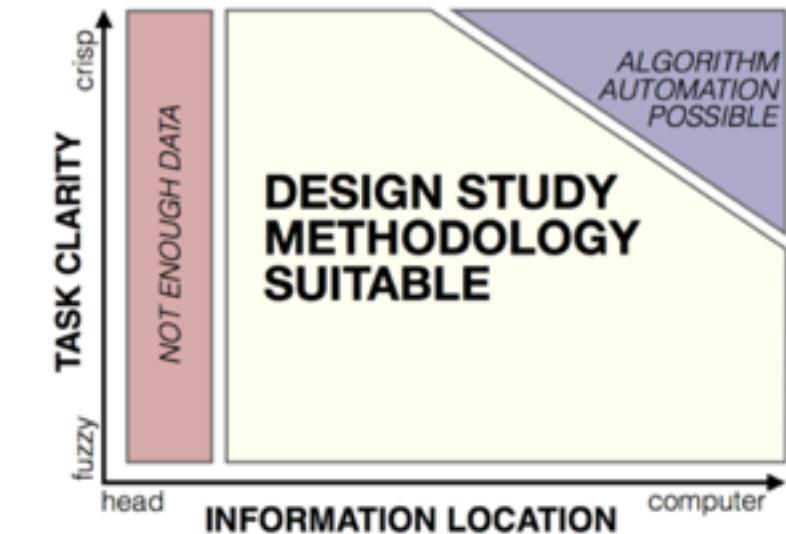
*Segal Design Institute, Northwestern University*

*8 April 2014, Chicago IL*

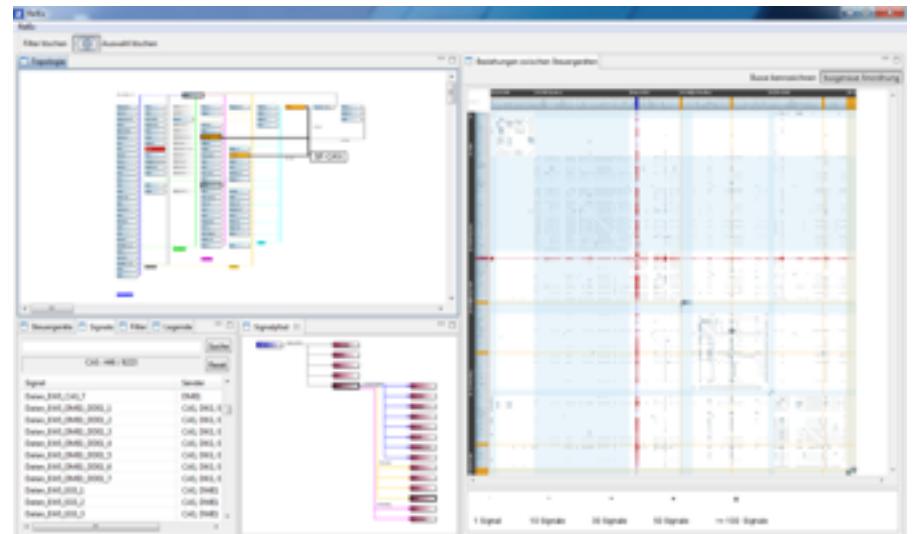
<http://www.cs.ubc.ca/~tmm/talks.html#chicago14>

# Trio

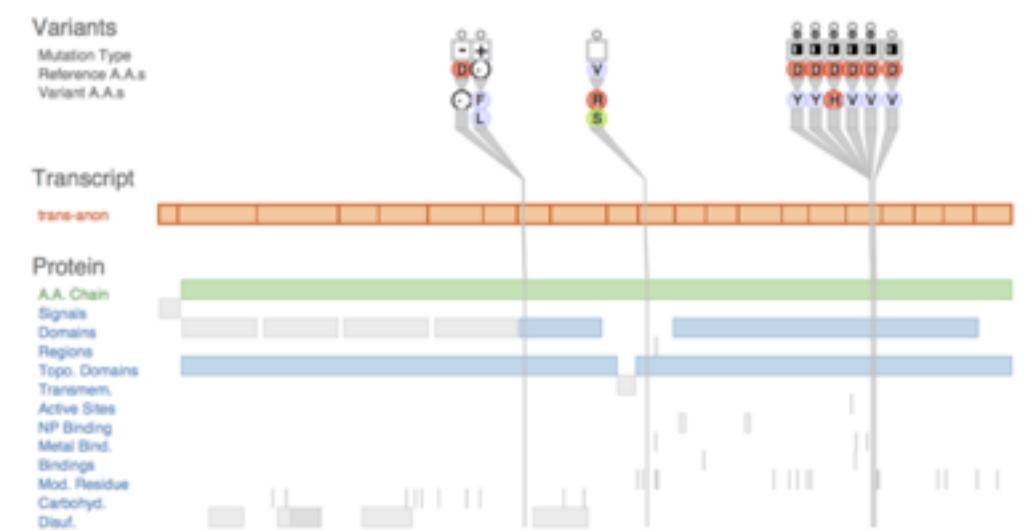
- Design Study Methodology
  - meta-paper: how to do design studies



- RelEx
  - overlay network optimization for in-car networks



- Variant View
  - sequence variant analysis in gene context



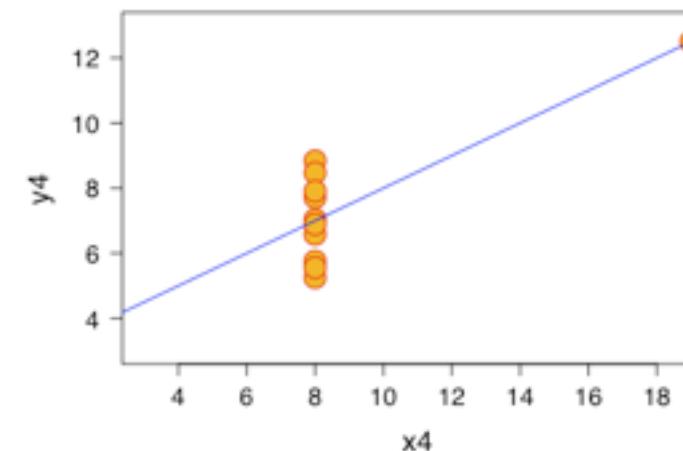
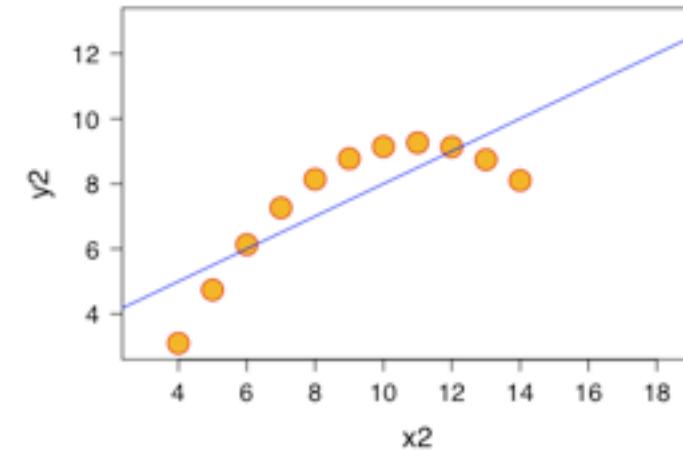
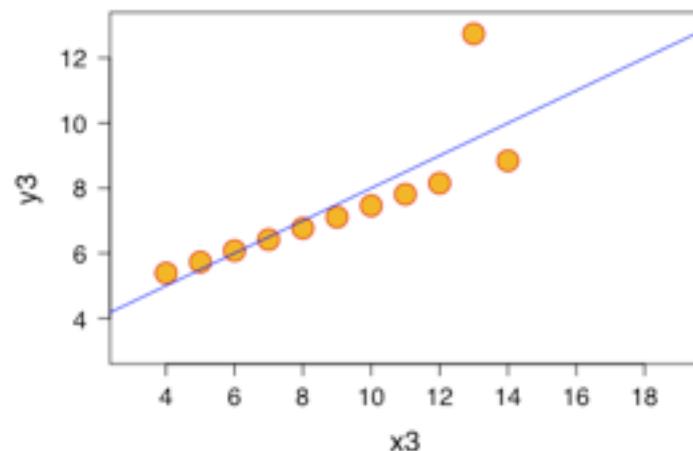
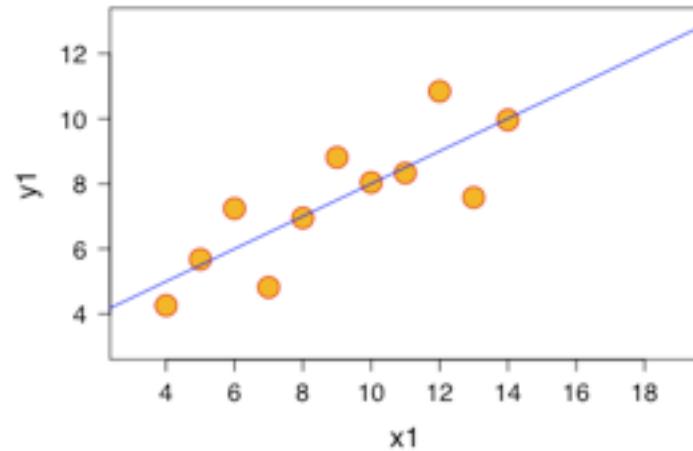
# Defining Visualization

**Computer-based visualization systems provide visual representations of datasets designed to help people carry out tasks more effectively.**

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**Computer-based visualization systems provide visual representations of datasets designed to help people carry out tasks more effectively.**

- human in the loop needs the details
  - doesn't know exactly what questions to ask in advance

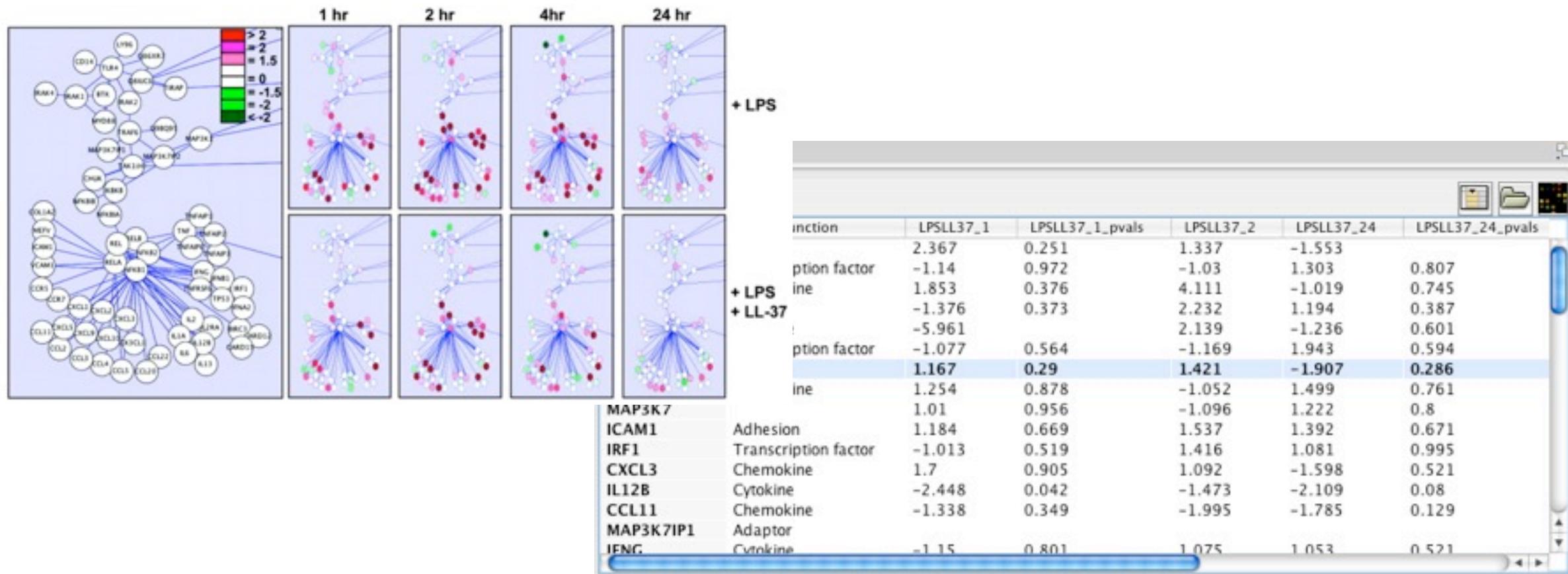


Identical statistics		
x mean	9.0	
x variance	10.0	
y mean	7.50	
y variance	3.75	
x/y correlation	0.816	

# Defining Visualization

**Computer-based visualization systems provide visual representations of datasets designed to help people carry out tasks more effectively.**

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- external representation: replace cognition with perception



# Defining Visualization

**Computer-based visualization systems provide visual representations of datasets designed to help people carry out tasks more effectively.**

- human in the loop needs the details
  - doesn't know exactly what questions to ask in advance
- external representation: perception vs cognition
- intended task

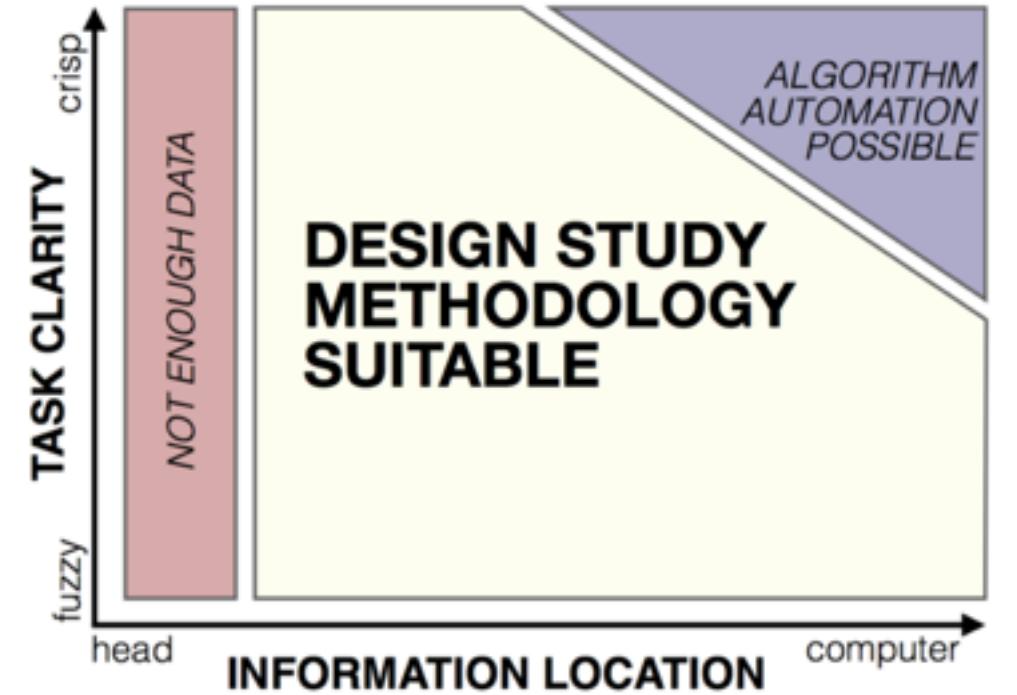
# Defining Visualization

**Computer-based visualization systems provide visual representations of datasets designed to help people carry out tasks more effectively.**

- human in the loop needs the details
  - doesn't know exactly what questions to ask in advance
- external representation: perception vs cognition
- intended task
- measureable definitions of effectiveness

more at:

Visualization Analysis and Design, Chapter 1.  
Munzner. AK Peters, 2014, to appear.



# Design Study Methodology

*Reflections from the Trenches and from the Stacks*

**joint work with:**

Michael Sedlmair, Miriah Meyer

<http://www.cs.ubc.ca/labs/imager/tr/2012/dsm/>

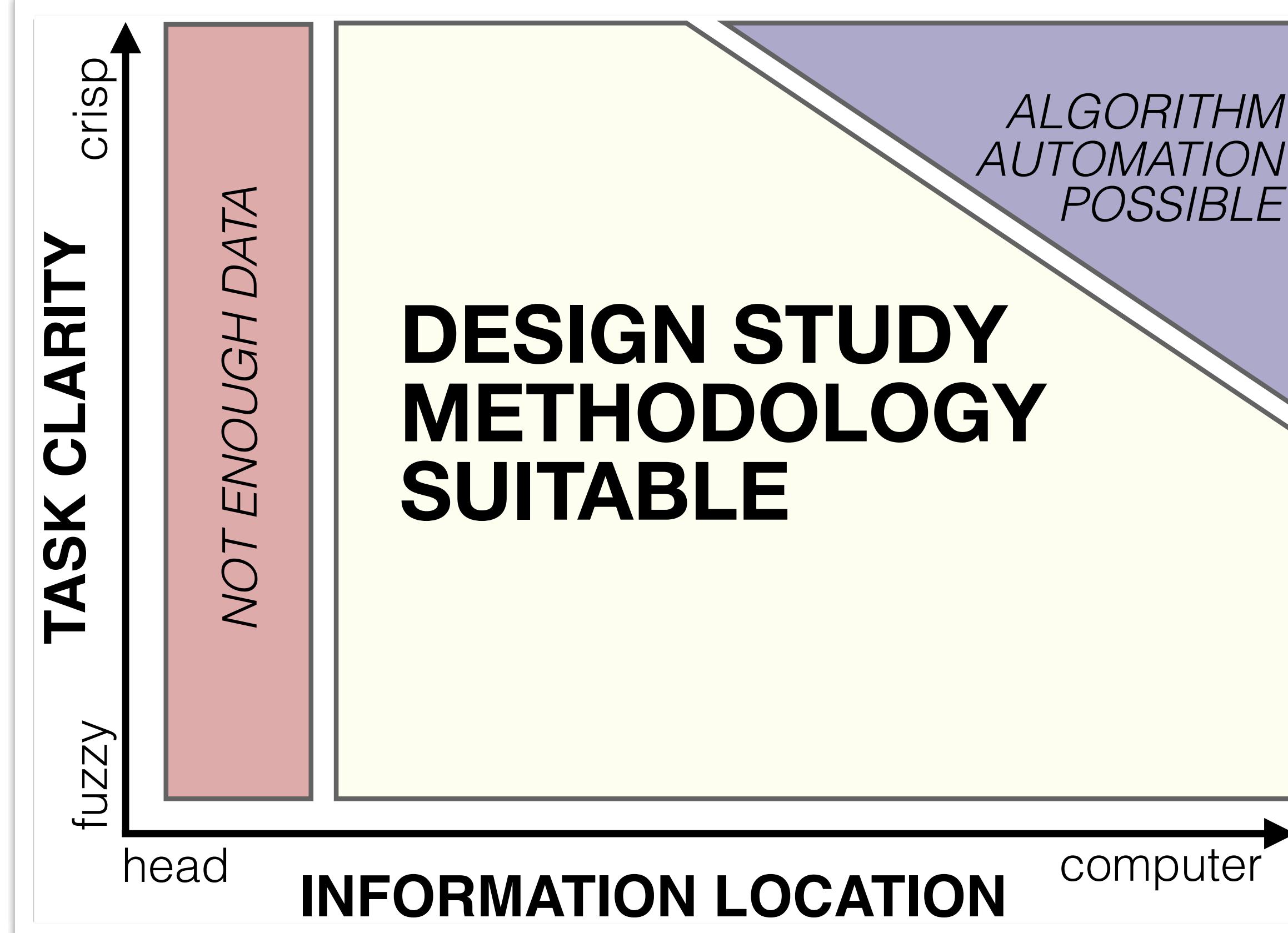
# Defining Design Study

- a specific **real-world** problem
  - real users and real data,
  - collaboration is (often) fundamental
- **design** a visualization system
  - implications: requirements, multiple ideas
- **validate** the design
  - at appropriate levels
- **reflect** about lessons learned
  - transferable research: improve design guidelines for vis in general
    - confirm, refine, reject, propose

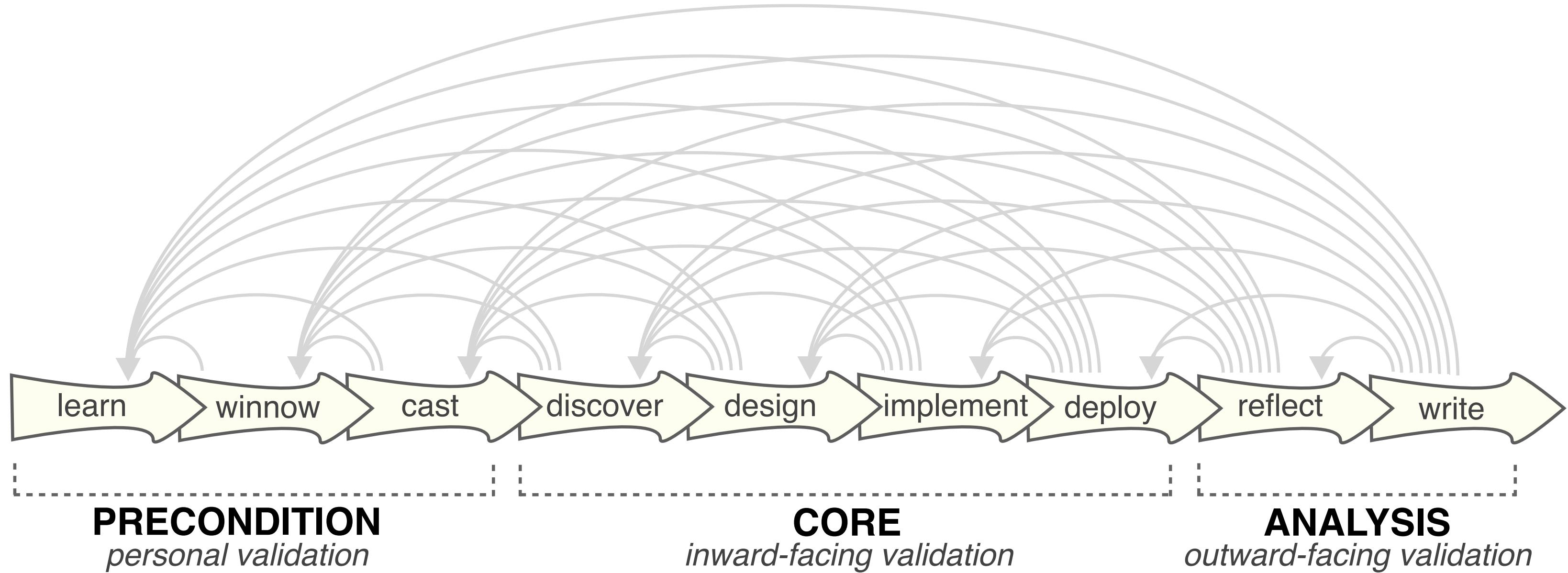
more at:  
A Nested Model of Visualization Design and Validation.  
Munzner. *IEEE TVCG* 15(6):921-928, 2009 (Proc. InfoVis 2009).

more at:  
The Nested Blocks and Guidelines Model.  
Meyer, Sedlmair, Quinan, Munzner. *Information Visualization Journal*, 2014,  
to appear.

# When To Do Design Studies

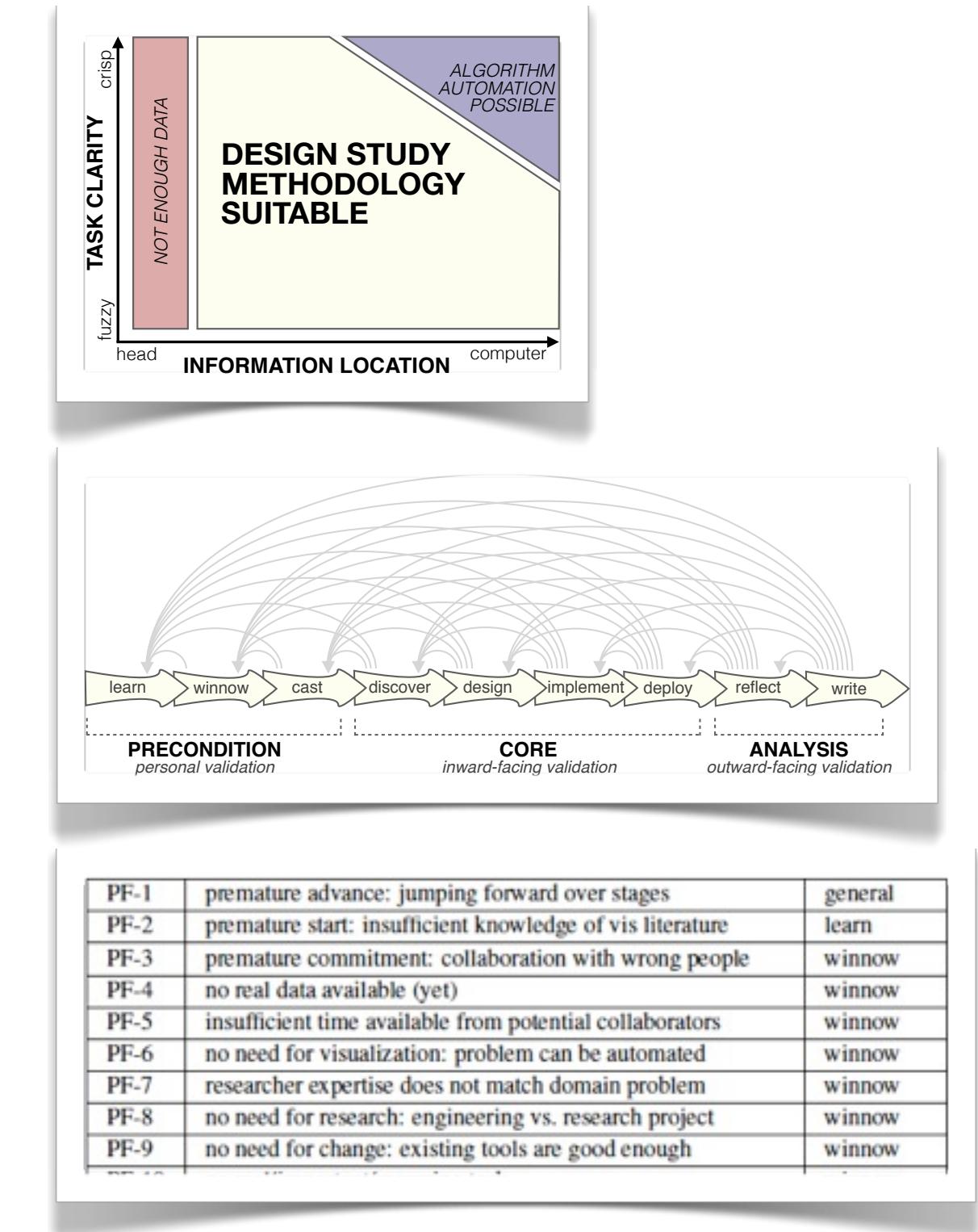


# Nine-Stage Framework



# How To Do Design Studies

- definitions
- 9-stage framework
- 32 pitfalls and how to avoid them



# Pitfall Example: Premature Publishing

algorithm innovation

design studies

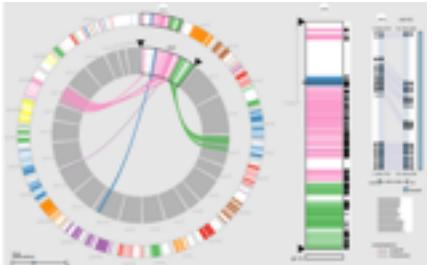
**Must be first!**



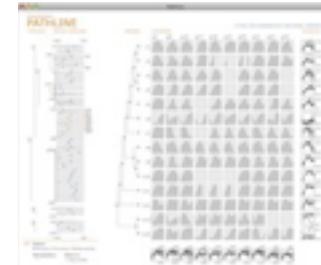
**Am I ready?**



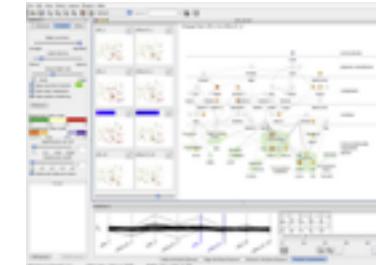
# Design Studies: Lessons learned after 21 of them



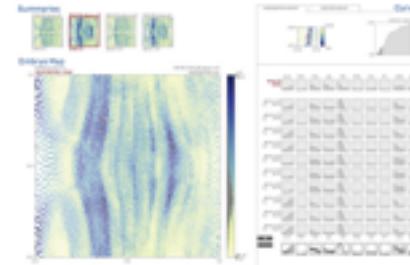
*MizBee*  
genomics



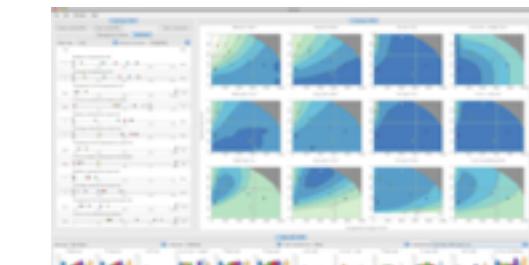
*Pathline*  
genomics



*Cerebral*  
genomics



*MulteeSum*  
genomics



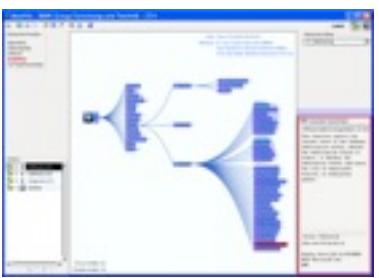
*Vismon*  
fisheries management



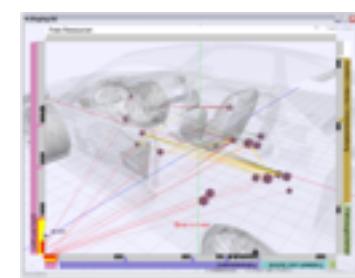
*QuestVis*  
sustainability



*WiKeVis*  
in-car networks



*MostVis*  
in-car networks



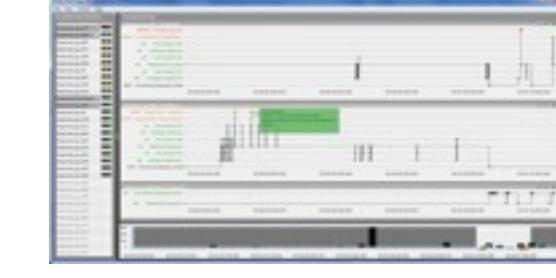
*Car-X-Ray*  
in-car networks



*ProgSpy2010*  
in-car networks



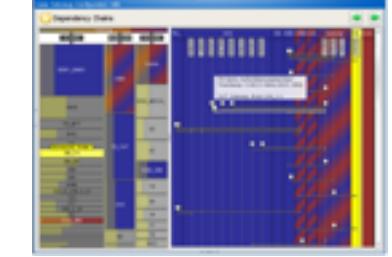
*ReIEx*  
in-car networks



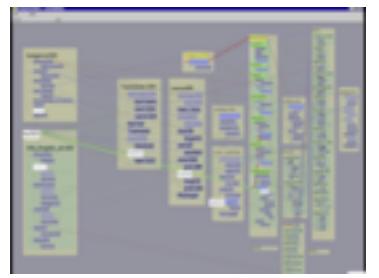
*Cardiogram*  
in-car networks



*AutobahnVis*  
in-car networks



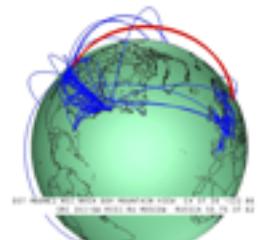
*VisTra*  
in-car networks



*Constellation*  
linguistics



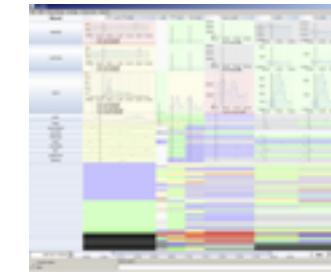
*LibVis*  
cultural heritage



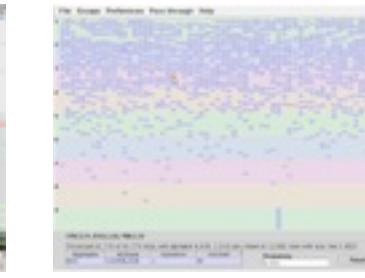
*Caidants*  
multicast



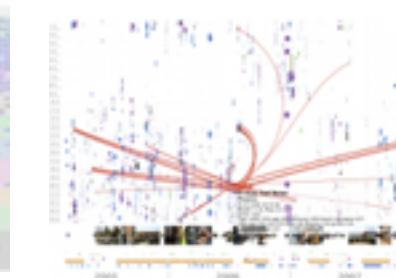
*SessionViewer*  
web log analysis



*LiveRAC*  
server hosting



*PowerSetViewer*  
data mining

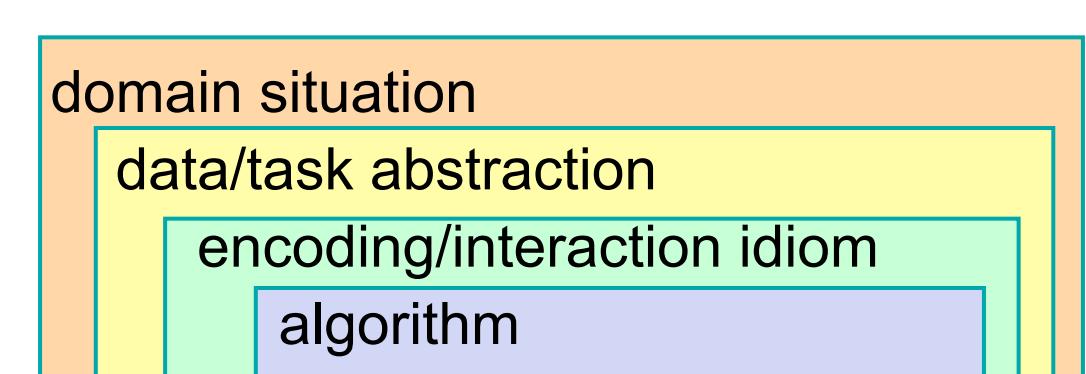


*LastHistory*  
music listening

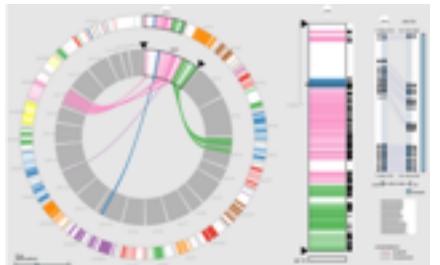
- commonality of representations cross-cuts domains!

# Abstractions and Idioms

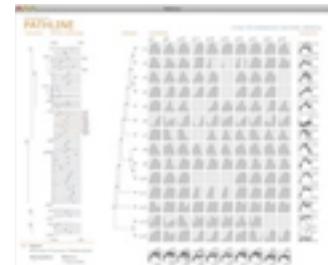
- abstractions
  - translate from specifics of domain to vocabulary of vis
    - task abstraction: **why** they're looking at it
    - data abstraction: **what** to draw
  - transform data into form useful for task at hand
    - don't just draw what you're given; decide what is the right thing!
- idioms
  - visual encoding idiom: **how** to draw
  - interaction idiom: **how** to manipulate
- focus today: two mappings
  - from domain to abstraction
  - from abstraction to idiom



# Today's Focus



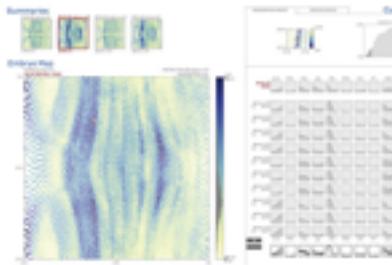
*MizBee*  
genomics



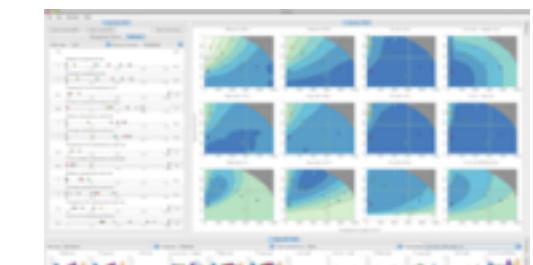
*Pathline*  
genomics



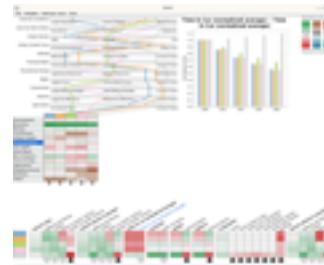
*Cerebral*  
genomics



*MulteeSum*  
genomics



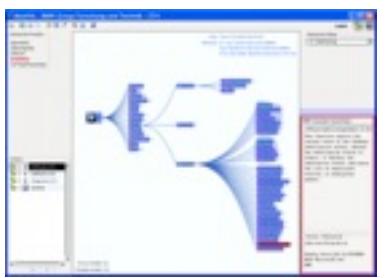
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fisheries management



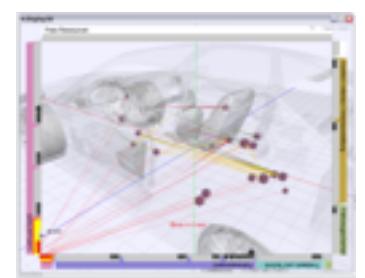
*QuestVis*  
sustainability



*WiKeVis*  
in-car networks



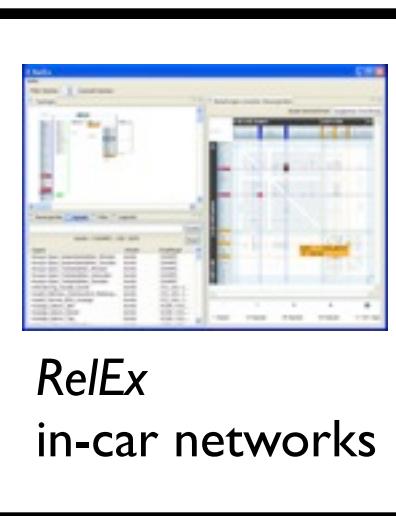
*MostVis*  
in-car networks



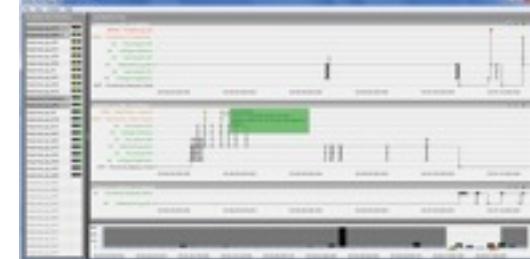
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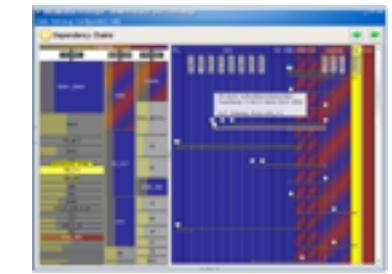
*RelEx*  
in-car networks



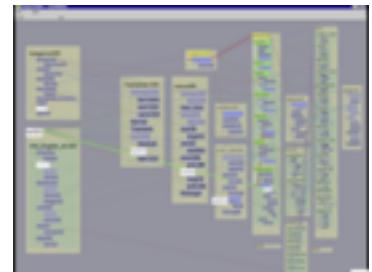
*Cardiogram*  
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*AutobahnVis*  
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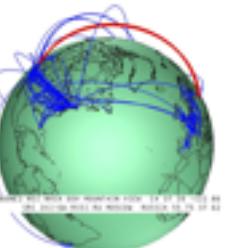
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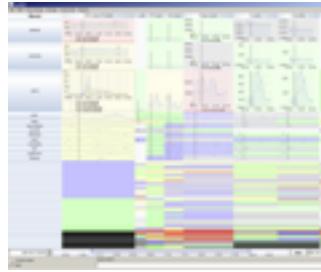
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cultural heritage



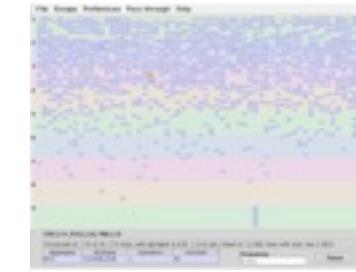
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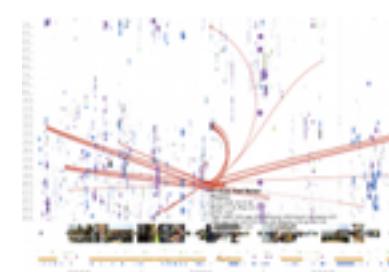
*SessionViewer*  
web log analysis



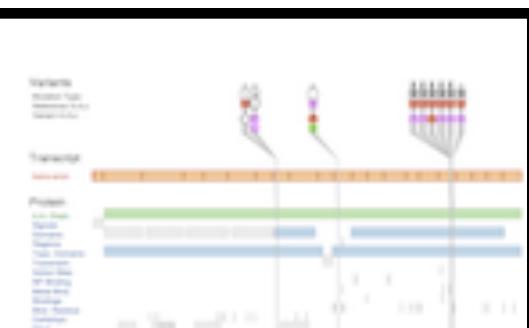
*LiveRAC*  
server hosting



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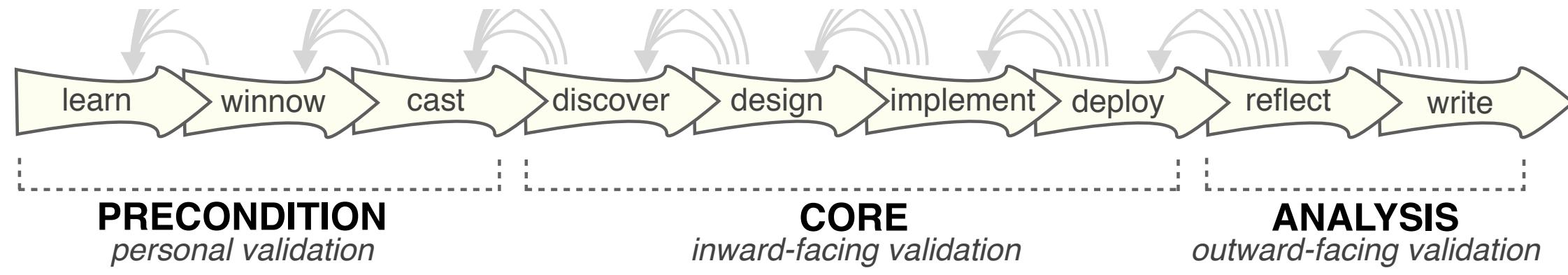
*LastHistory*  
music listening



*VariantView*  
genomics

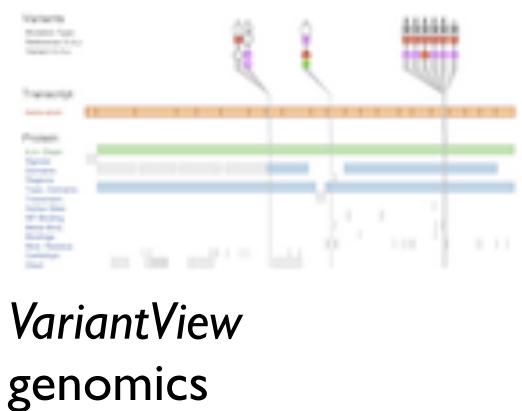
# Themes

- task and data abstraction
  - both cases: complex and tricky
  - clear description in final talk/paper is end of a long, long road
    - writing as research: refine during reflection even after vis tool is finalized...



RelEx  
in-car networks

- visual encoding and interaction idioms
  - RelEx: reduce memory load with interaction
  - VariantView: reduce interaction load with better visual encoding



VariantView  
genomics

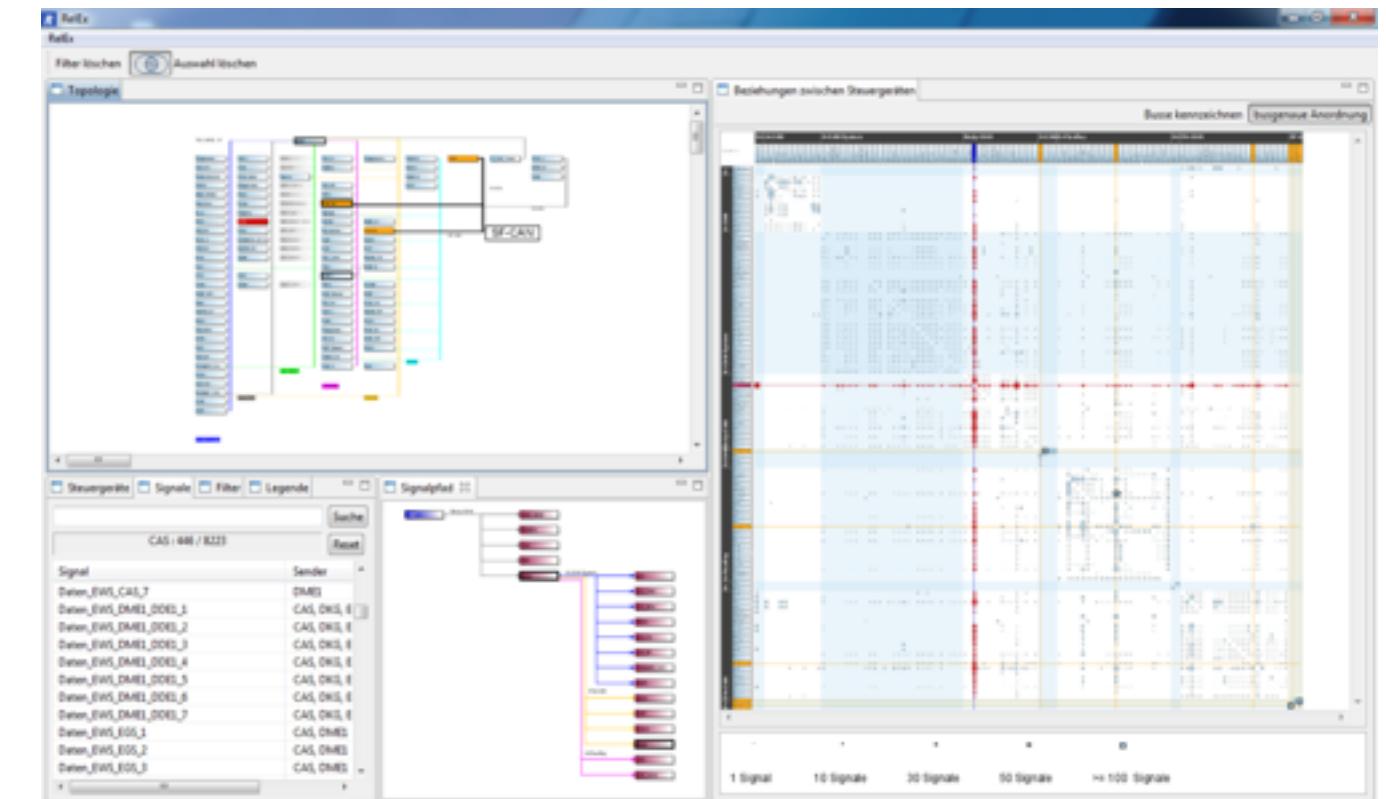
# ReEx

## *Visualization for Actively Changing Overlay Network Specifications*

**joint work with:**

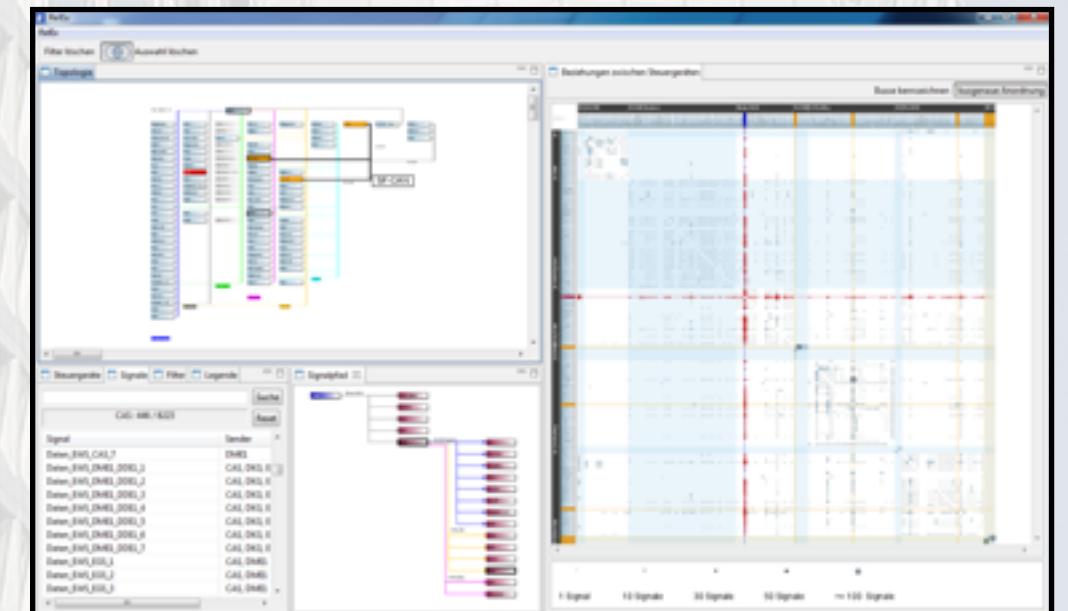
Michael Sedlmair, Annika Frank, Andreas Butz

<http://www.cs.ubc.ca/labs/imager/tr/2012/relex/>



RelEx: Visualization for Actively Changing Overlay Network Specifications.  
Sedlmair, Frank, Butz, Munzner. IEEE TVCG 18(12): 2729-2738, 2012 (Proc. InfoVis 2012).

# Domain: In-car network engineering



# Abstractions

# DATA

# In-car Electronics



# Data Abstraction: 3 Networks

- **physical network**

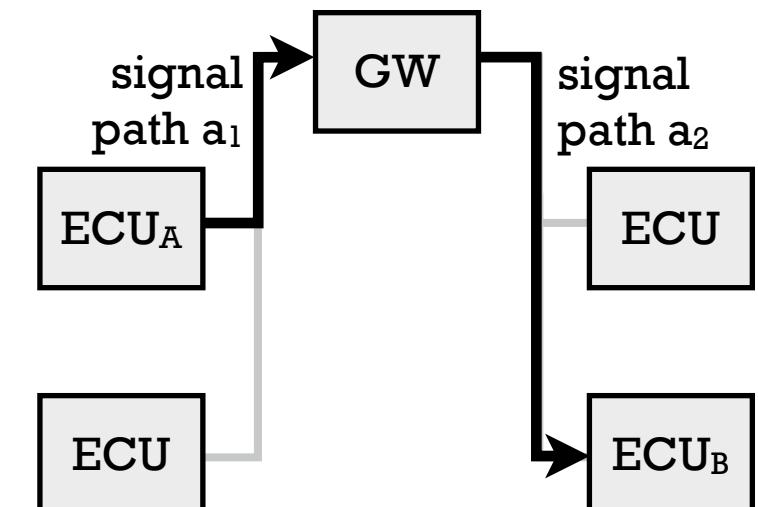
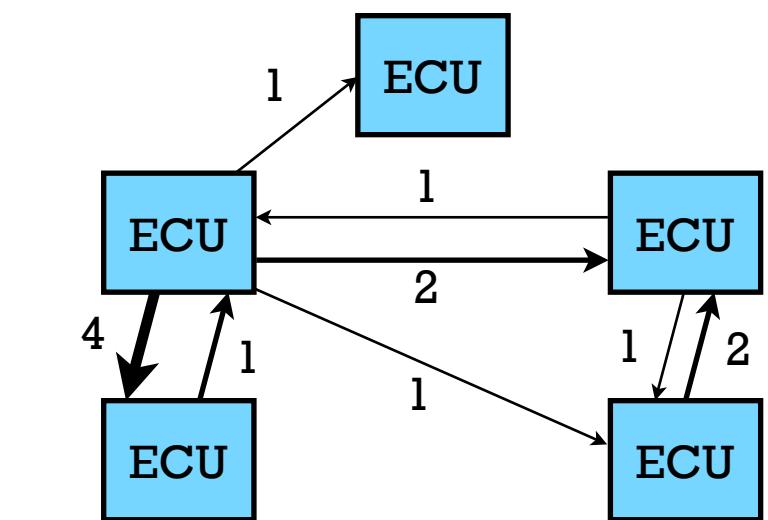
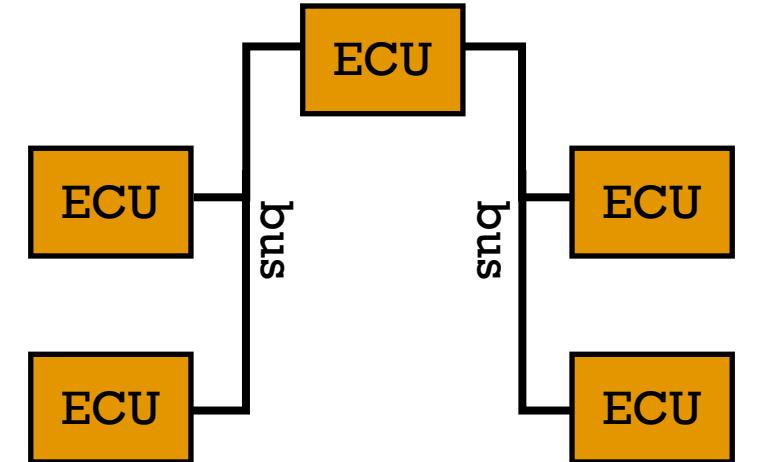
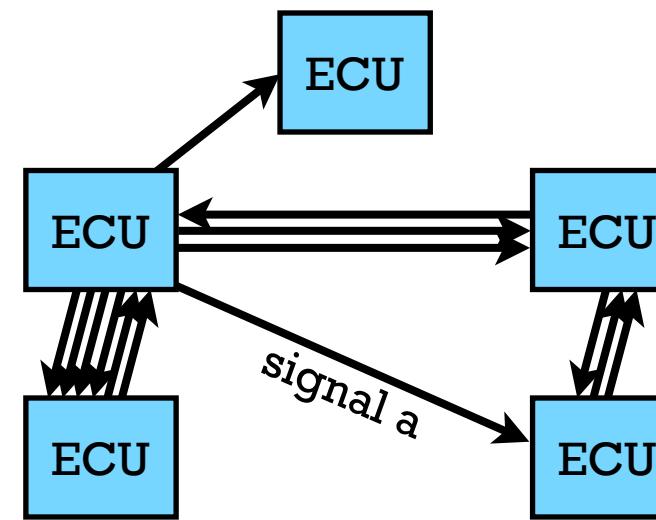
- 100 nodes: *Electronic Control Units*
- 10-15 hyperedges: *bus systems*
- hardware engineers

- **logical network**

- same nodes
- 10,000 multigraph edges: *signals*
- 1,000 weighted edges: *signal counts*
- software engineers

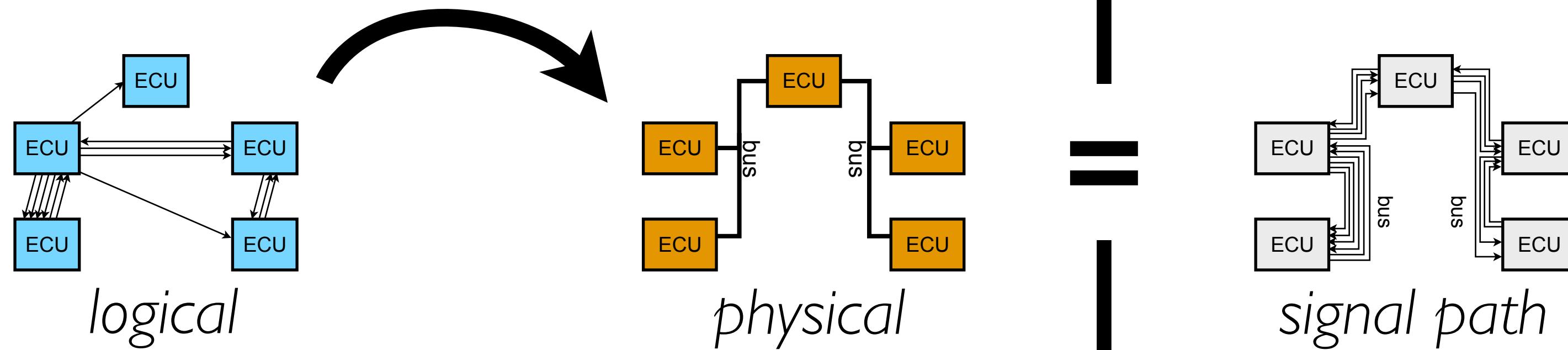
- **overlay network**

- maps logical onto physical
- 30,000 edges: *signal paths*
- target engineers



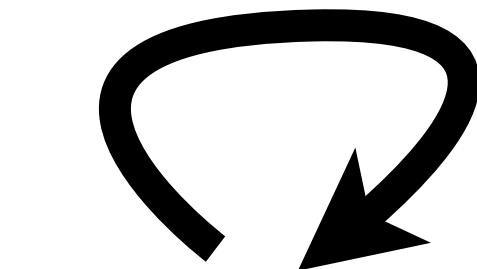
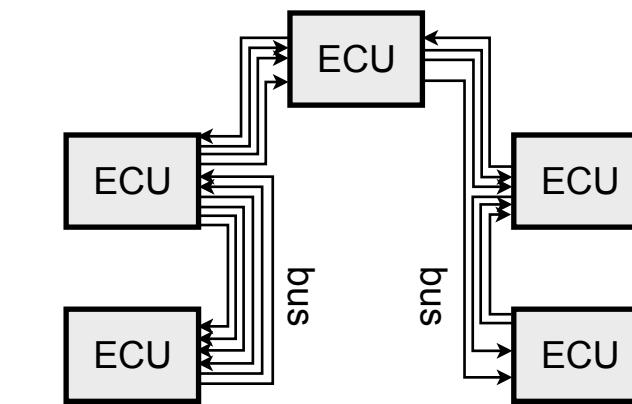
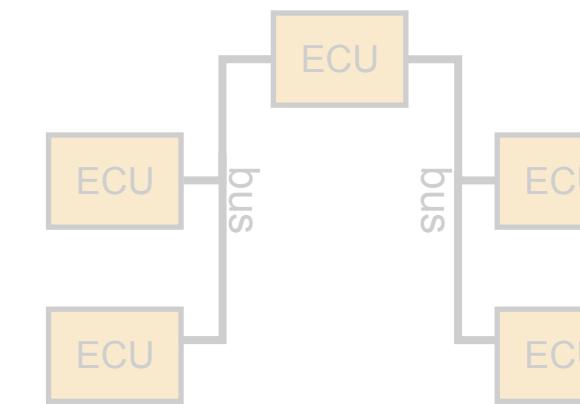
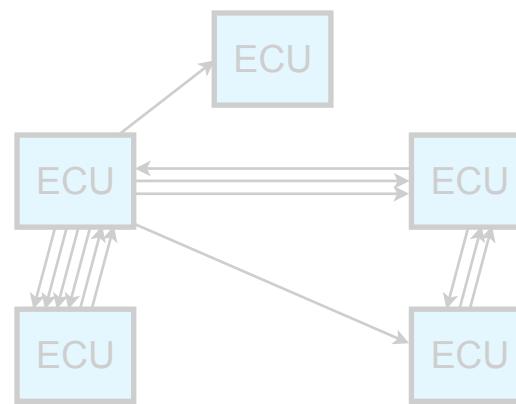
# Task Abstraction: Mapping

- specify overlay network that maps logical onto physical



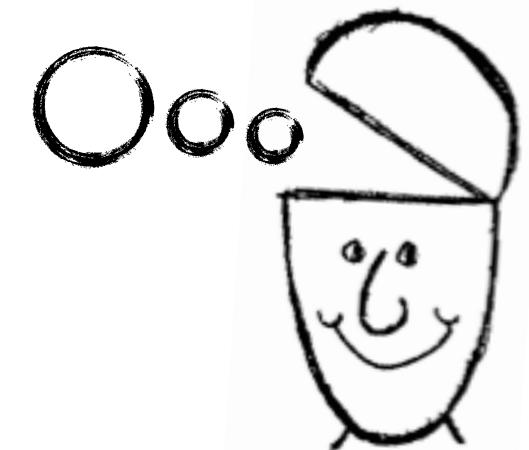
# Task Abstraction: Optimizing

- traffic optimization



## Many constraints

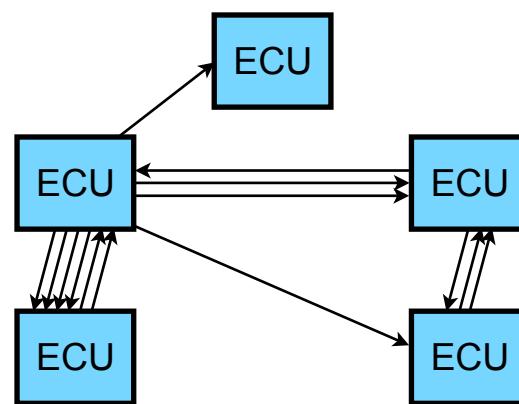
bandwidth ... delay/real time ...  
path length ... load balance ...  
reliability ... money ...



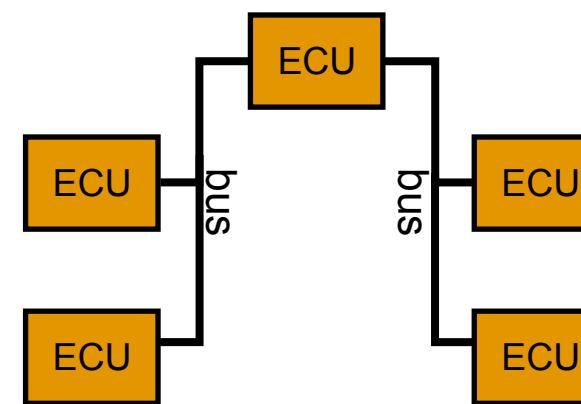
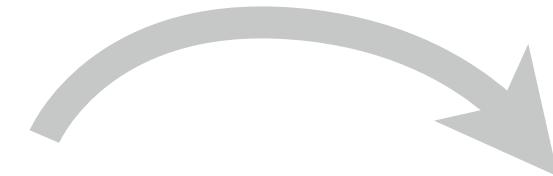
-- engineer, BMW --

# Task Abstraction: Changing

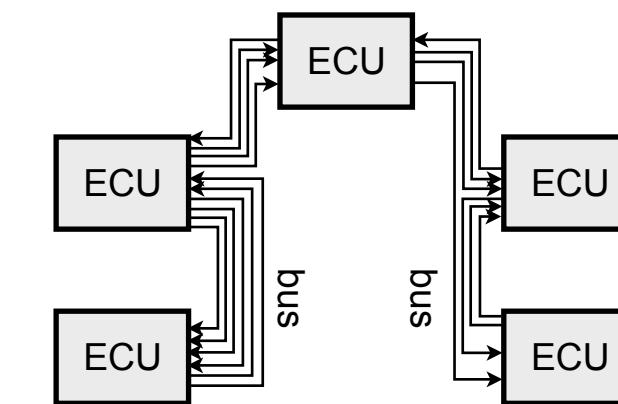
- external change requests



*logical*



*physical*



*signal path*



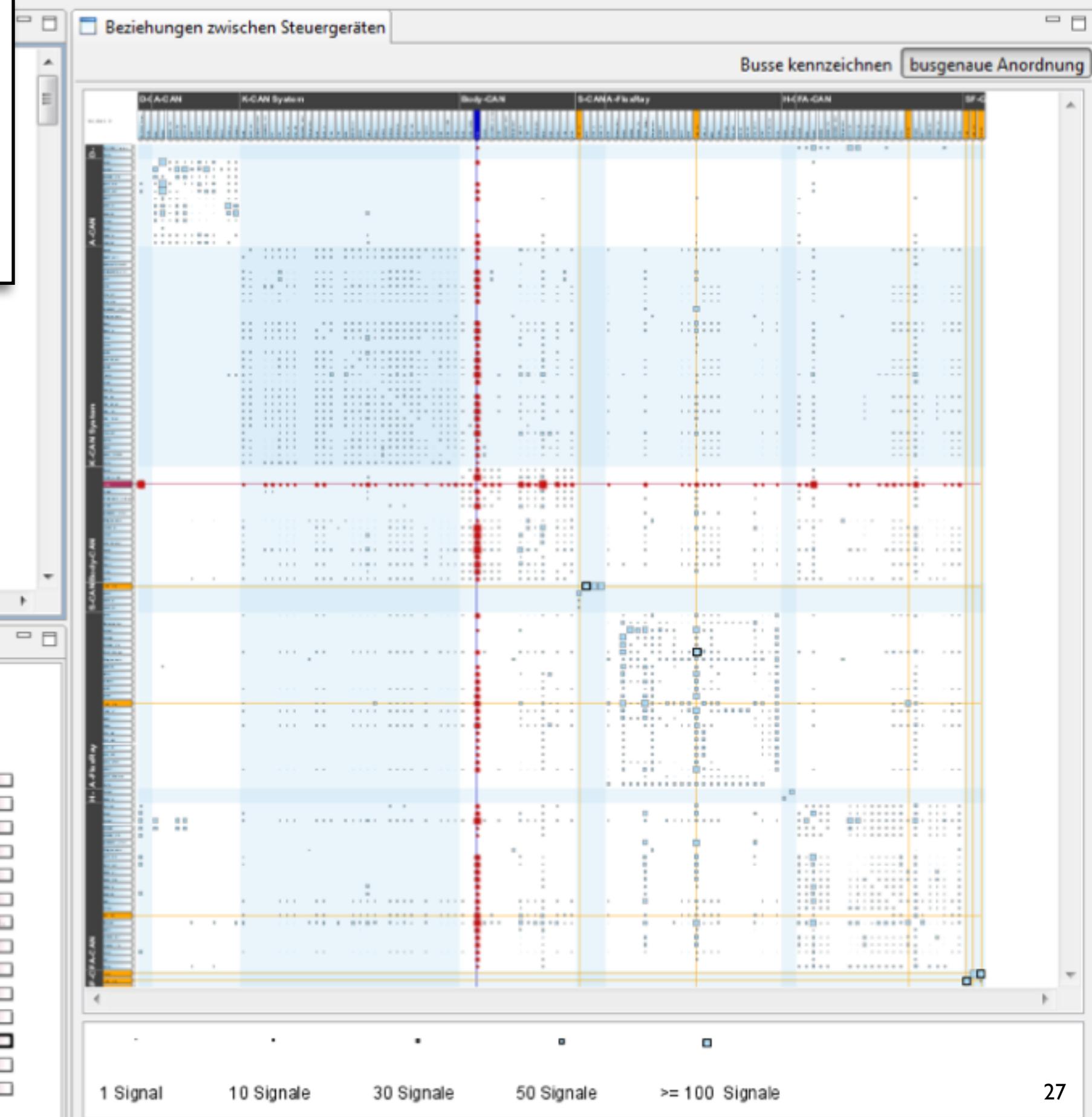
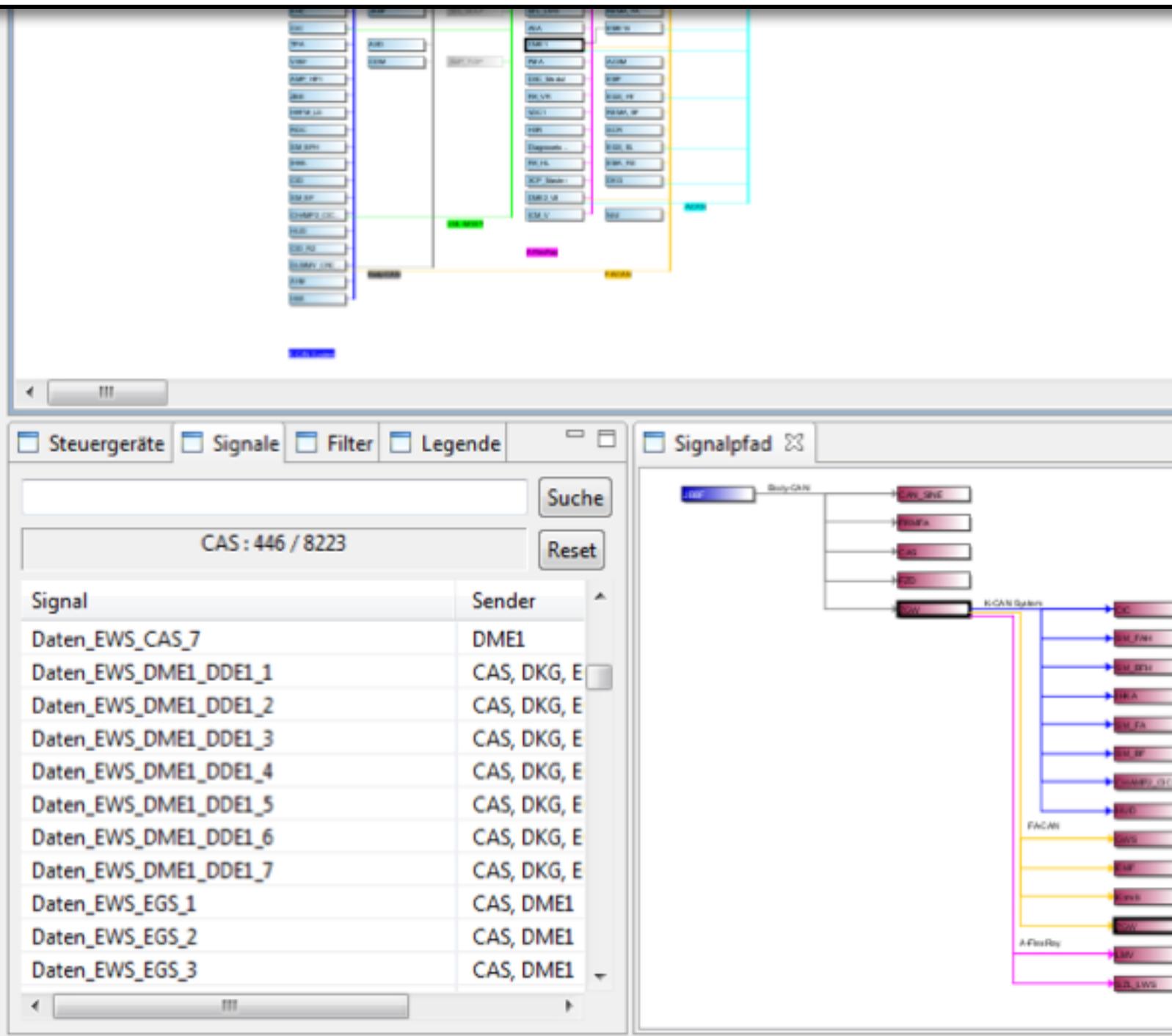
**Change**

*(trivial requests might lead to complex changes)*

# Idioms

# RELEX:

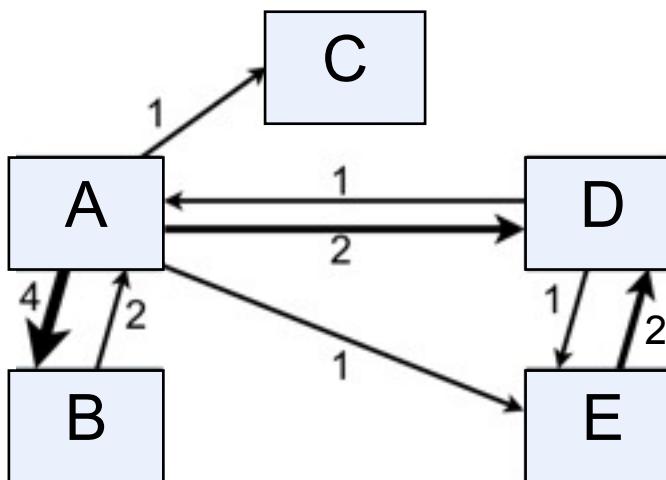
# Relation Explorer



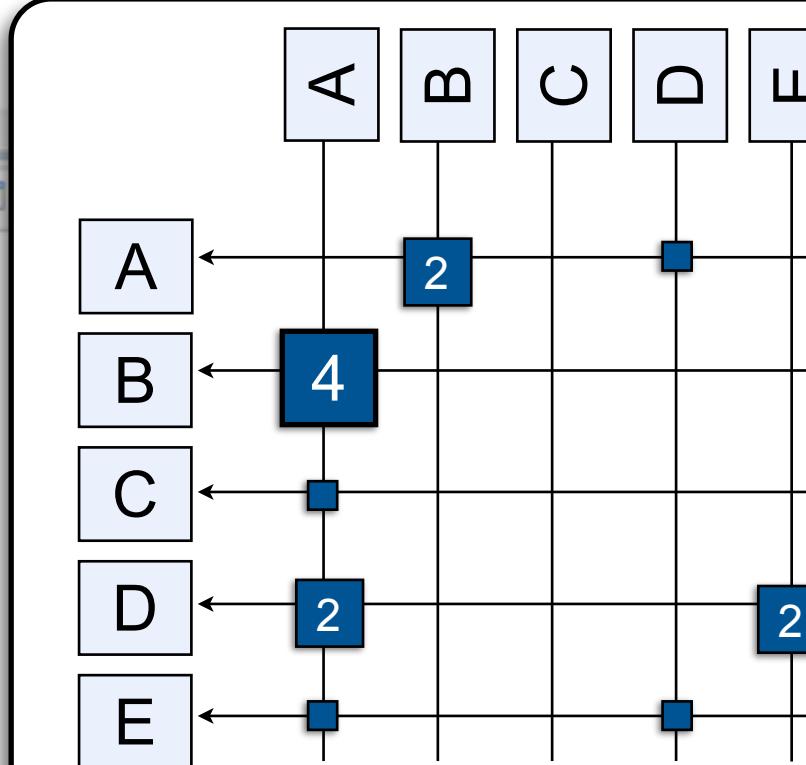
# Vis Guideline [Ghoniem 2005]

## Matrix for dense network data

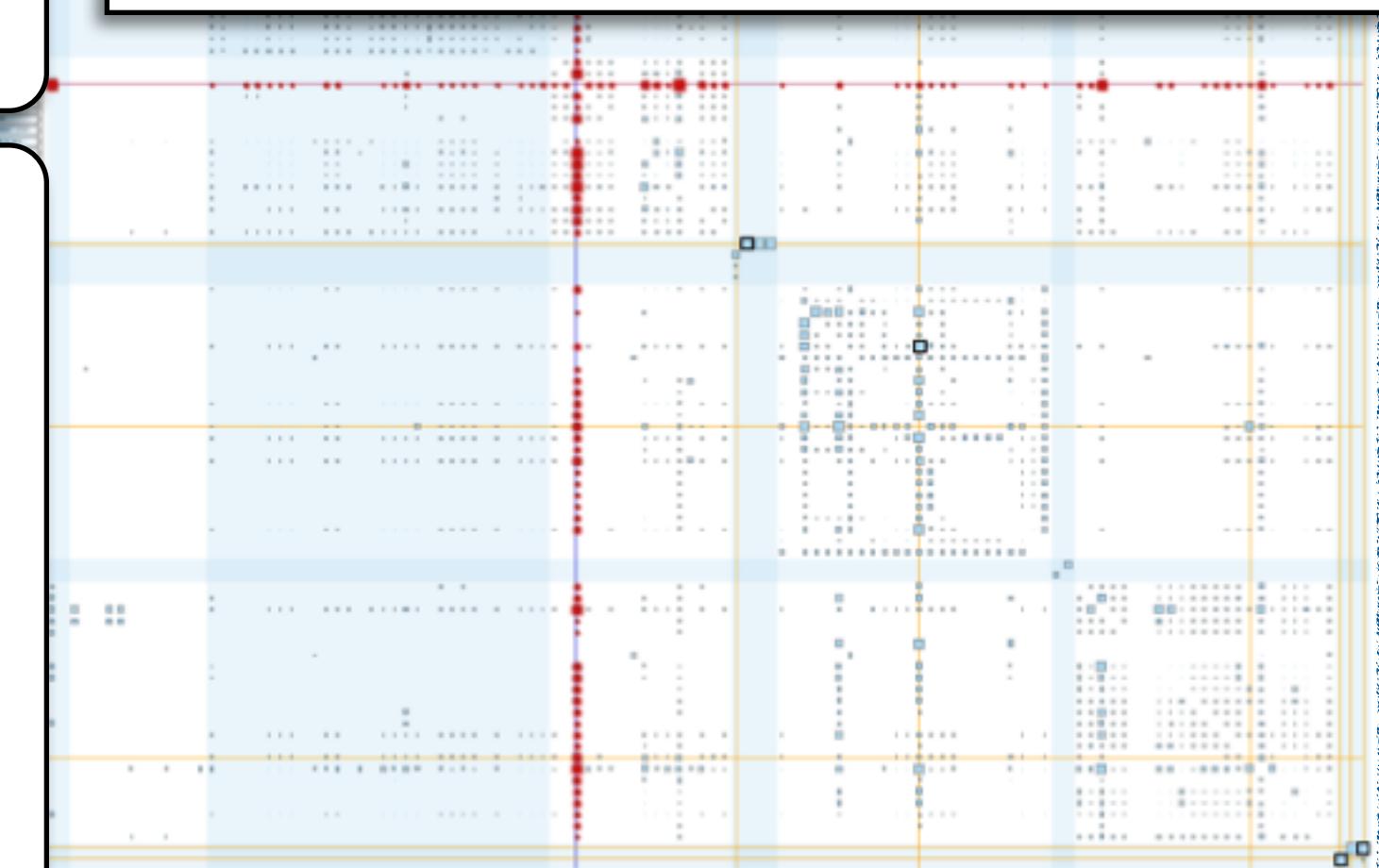
### SIGNAL COUNT NETWORK



visual encoding:  
size-coded matrix



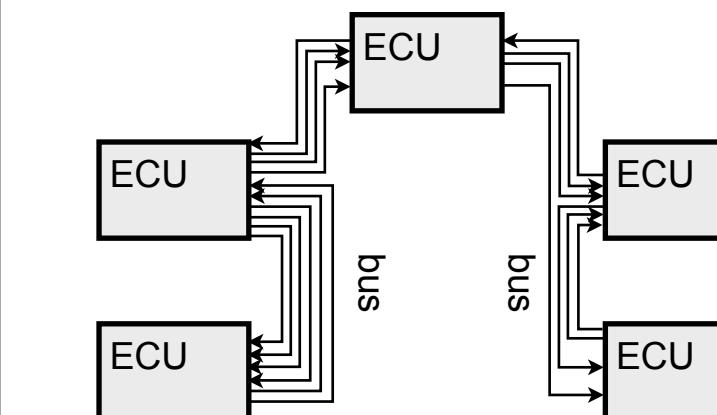
### Logical Network View: Overview



# Vis Guideline [Ghoniem 2005]

## ◀ for path following tasks

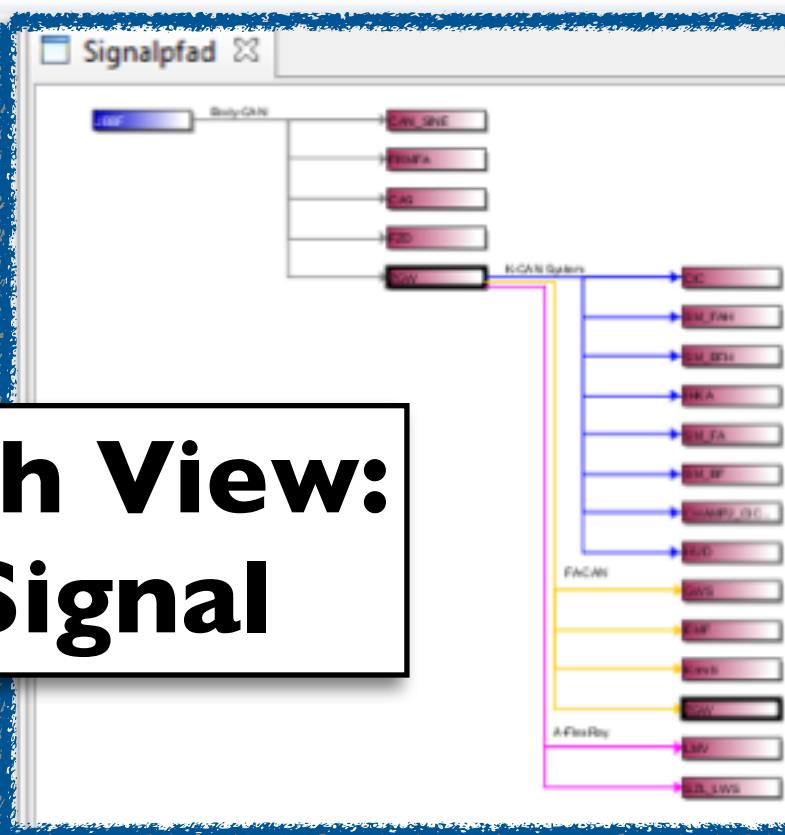
# SIGNAL PATH NETWORK



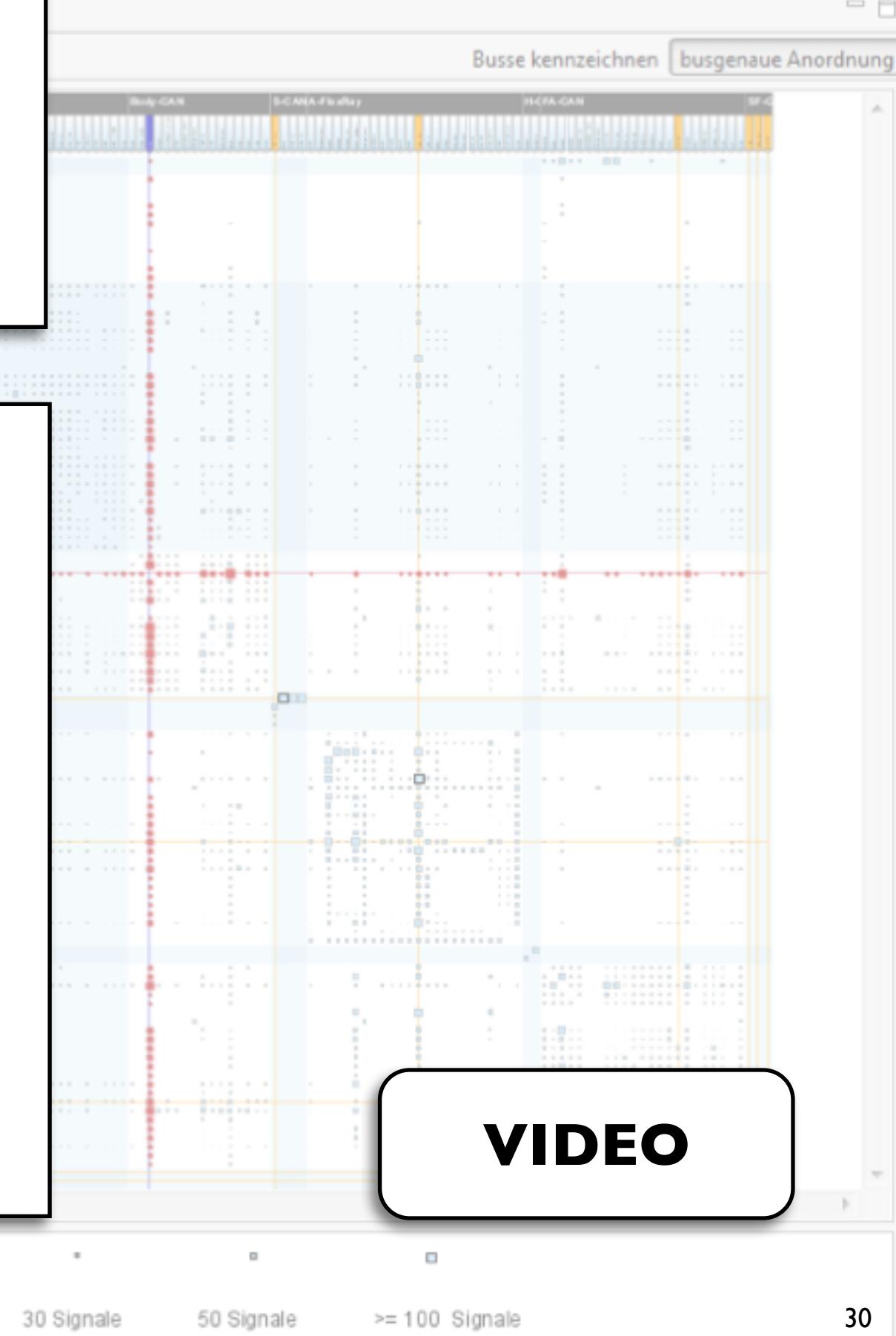
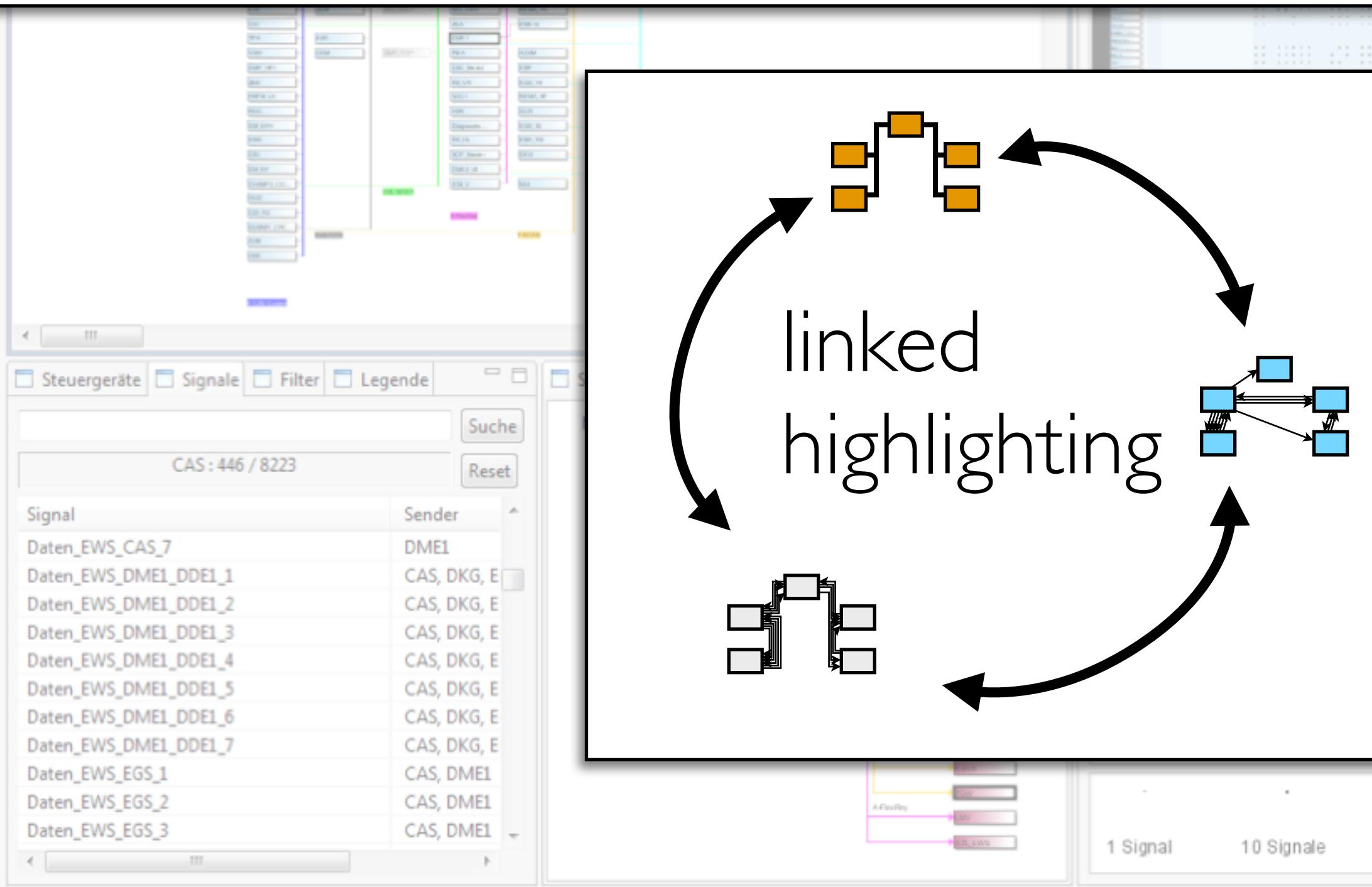
# filtered by signa

# **Signal Path View**

## **Selected Signal**

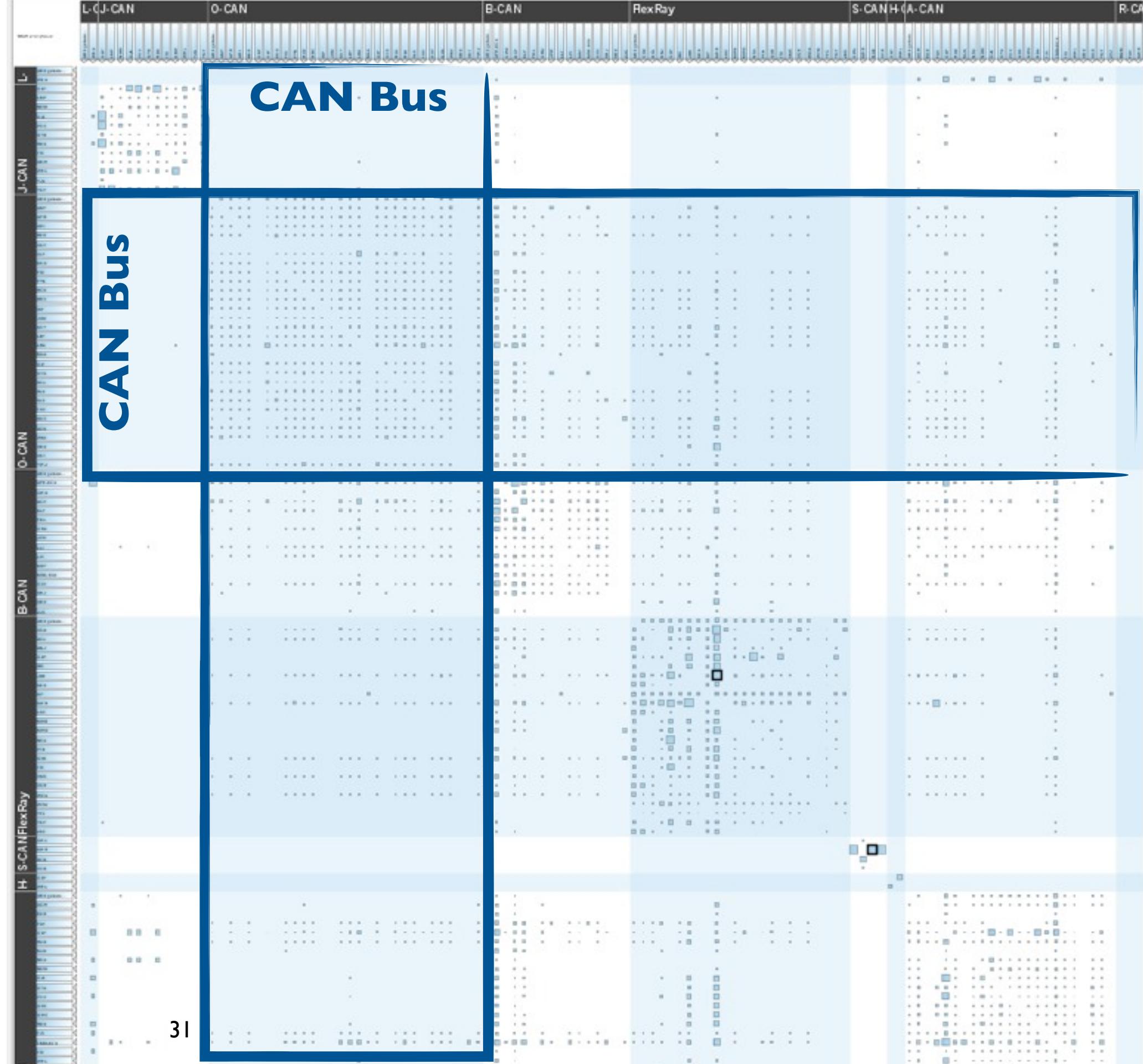


# INTERACTION IDIOM: Cross-Network Relations



**VIDEO**

# INTERESTS Bus communication patterns

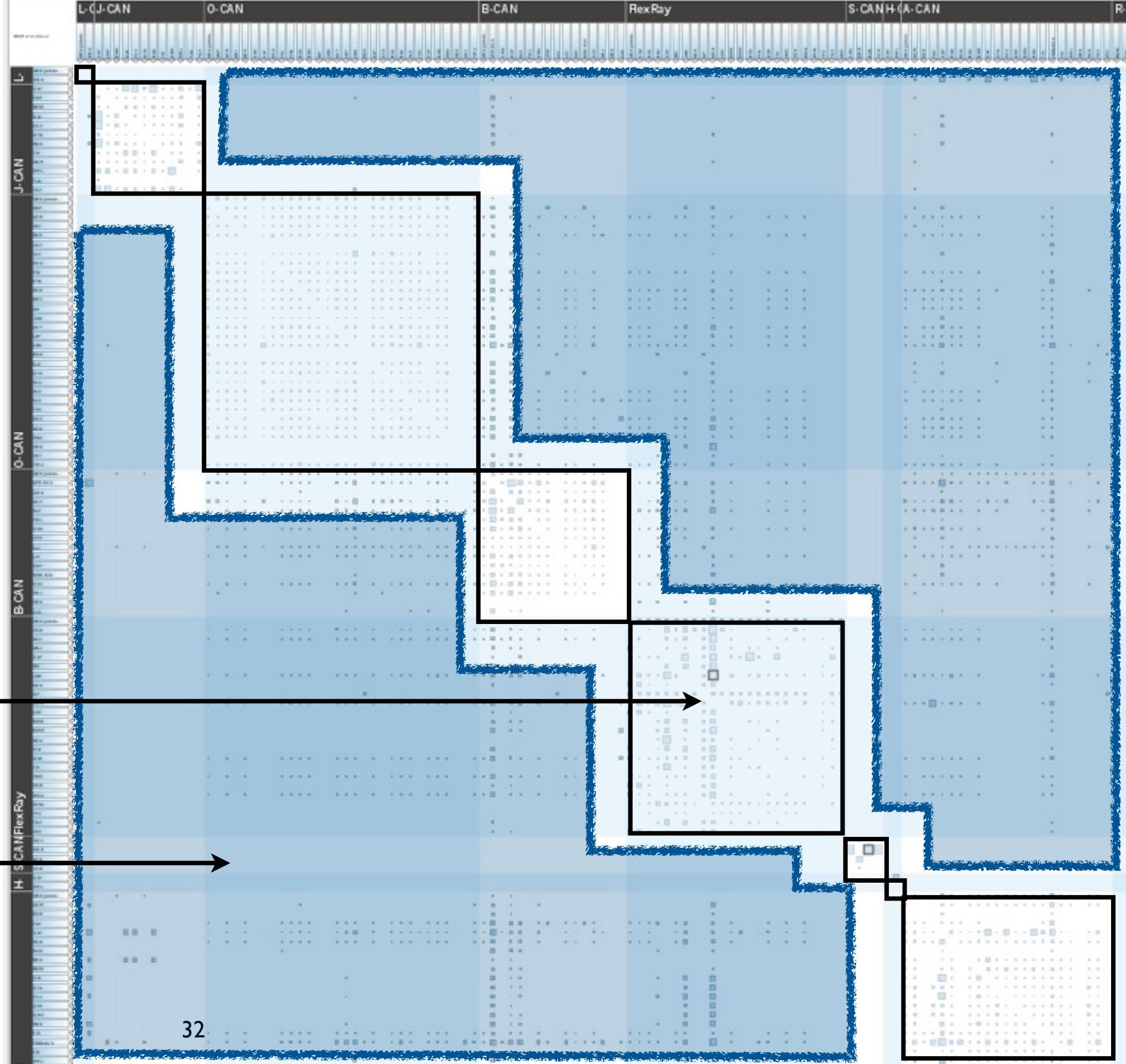


# INTERESTS

## Bus communication patterns

**Within-bus**

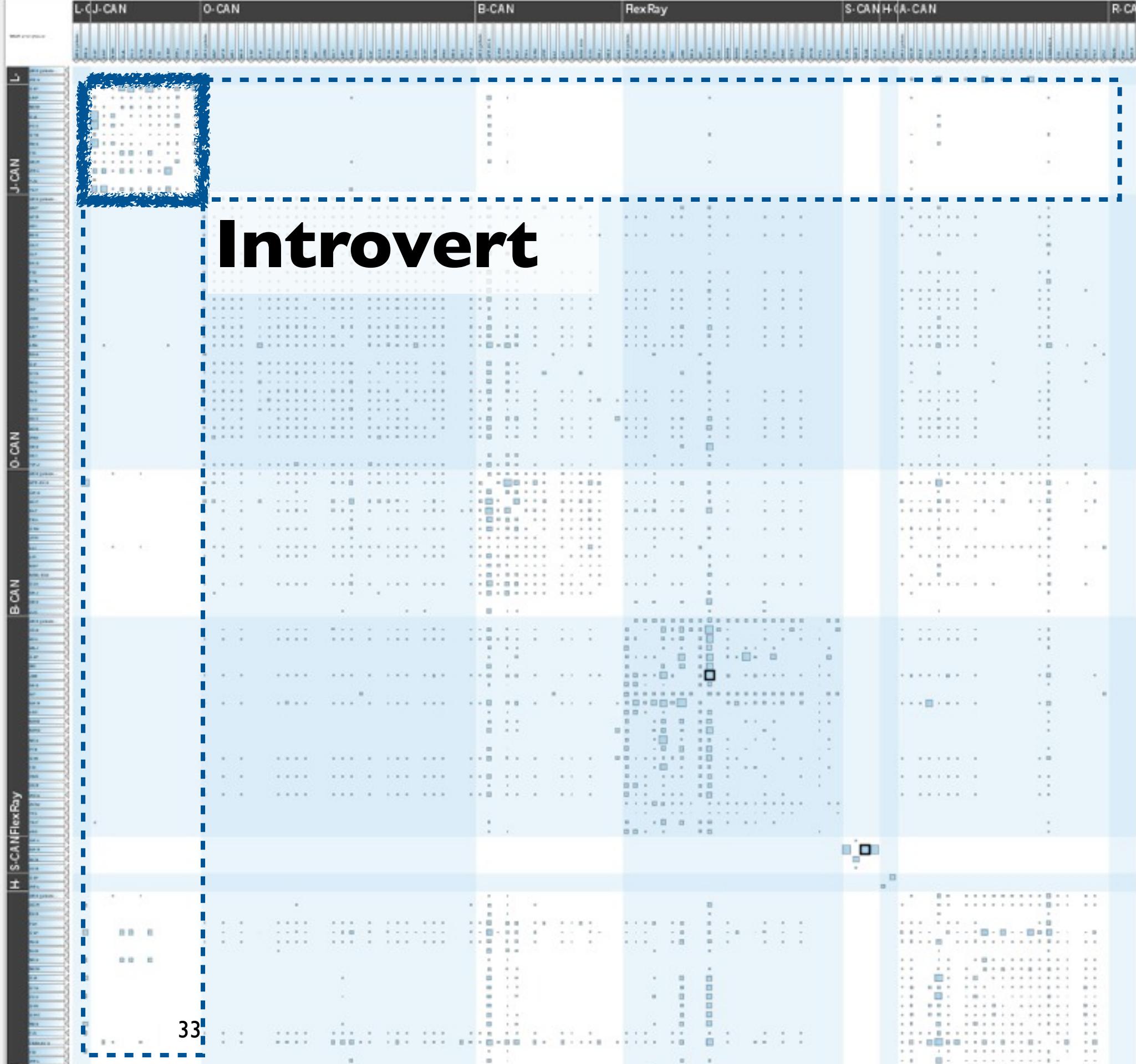
**Between-bus**



# INTERESTS

## Bus communication patterns

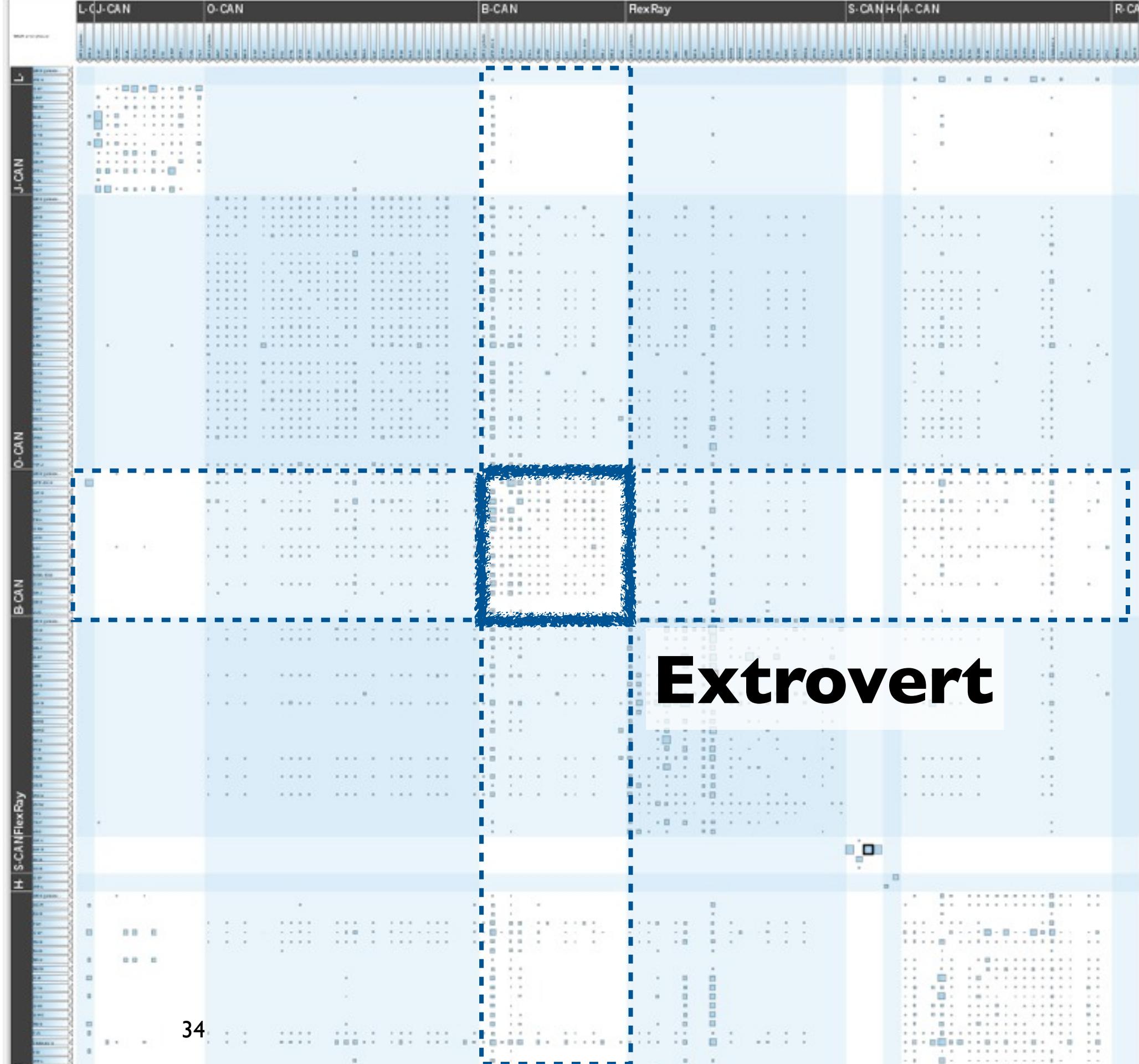
introvert  
vs.  
extrovert



# INTERESTS

## Bus communication patterns

introvert  
vs.  
extrovert



# Methods

# Phase I: Discover

3 months



- embedded within BMW
  - phases I, 2, 3
- contextual inquiry
- abstracting
- deriving design requirements

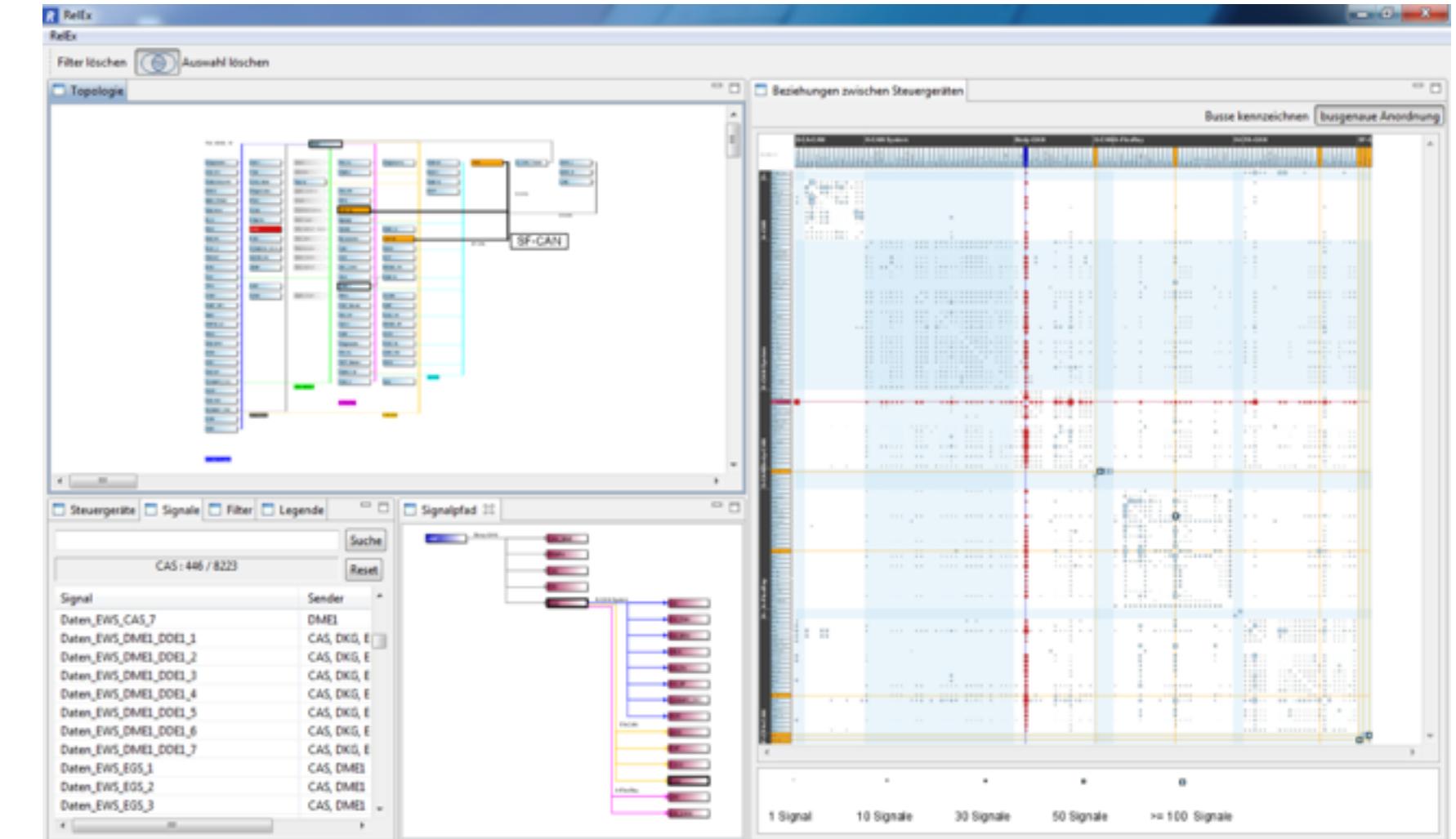


# Phase 2: Design, implement, deploy

4 months



- iterative paper prototyping
- agile software development
  - 3 lead users (engineers)
  - 6 deployed releases
- usability engineering
  - domain experts
  - HCI students



## Phase 3: Summative evaluation

2 months



- field study
  - 7 engineers
  - 5 weeks
- think aloud study
  - 10 engineers
  - ~1 hour each session
- adoption
  - 15+ users, 3 months post-study



# Phase 4: Reflect and write

3 months



- revisit abstractions
- relate to other design studies
- write up

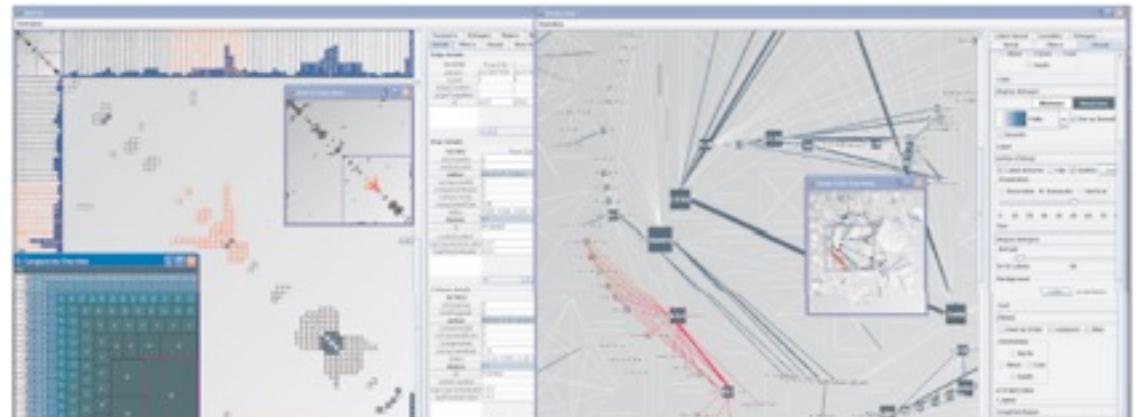


# Abstraction Innovation

# Previous Work

## Focus on social network analysis

- radically different task and data abstractions



MatrixExplorer



SocialAction



Honeycomb

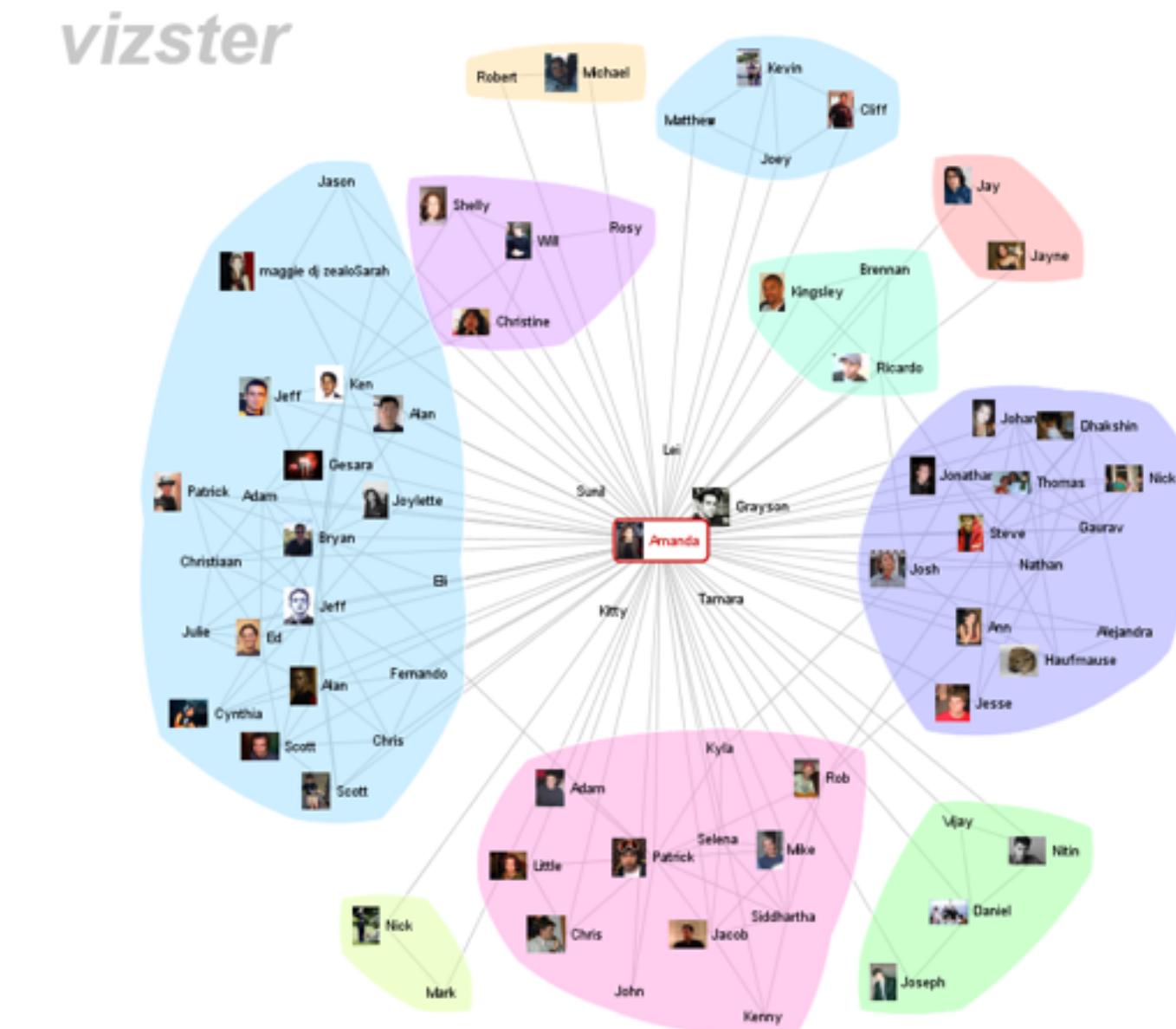


vizster

# Task Abstraction

# Social Network Analysis Domain

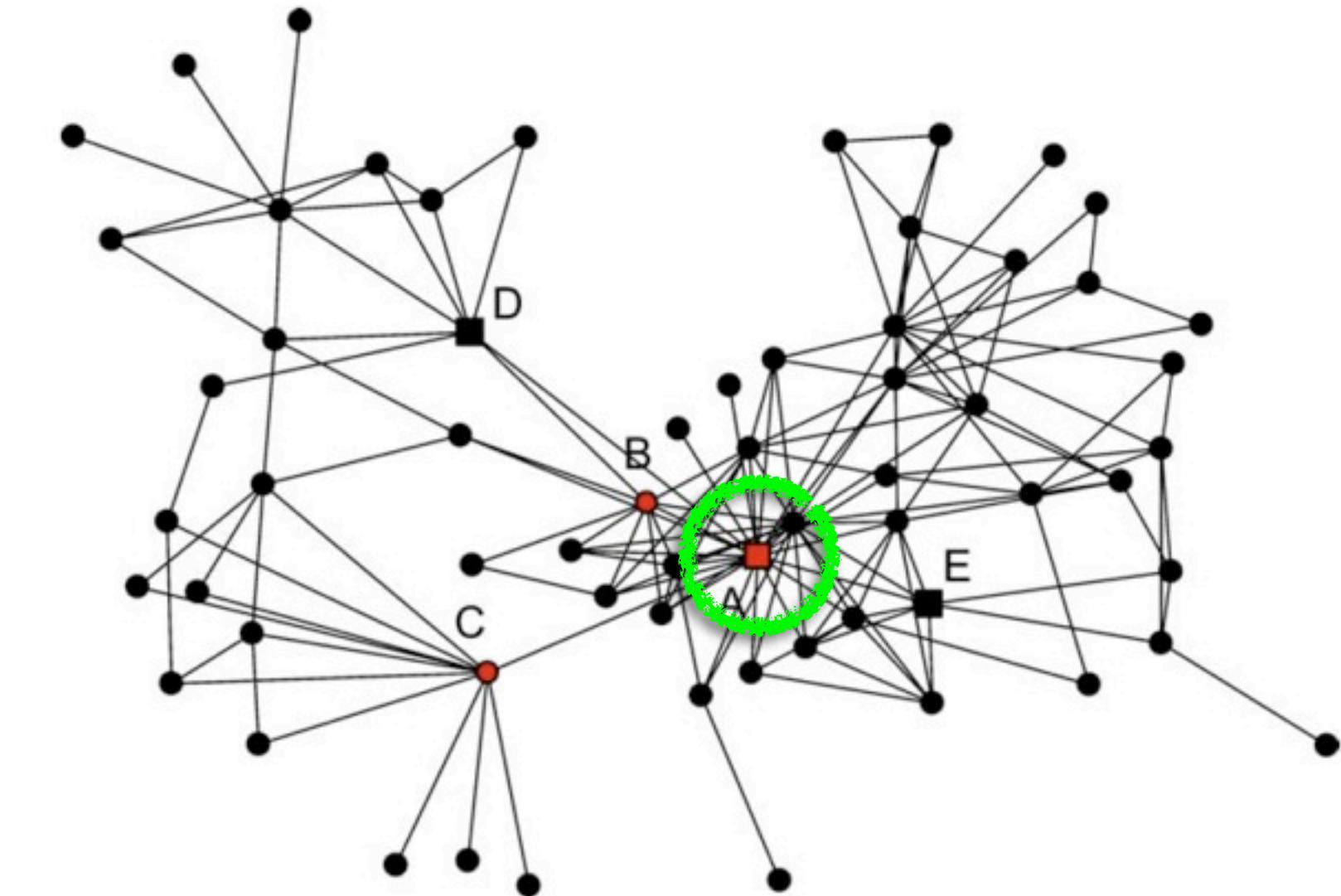
- find clusters



# Task Abstraction

## Social Network Analysis Domain

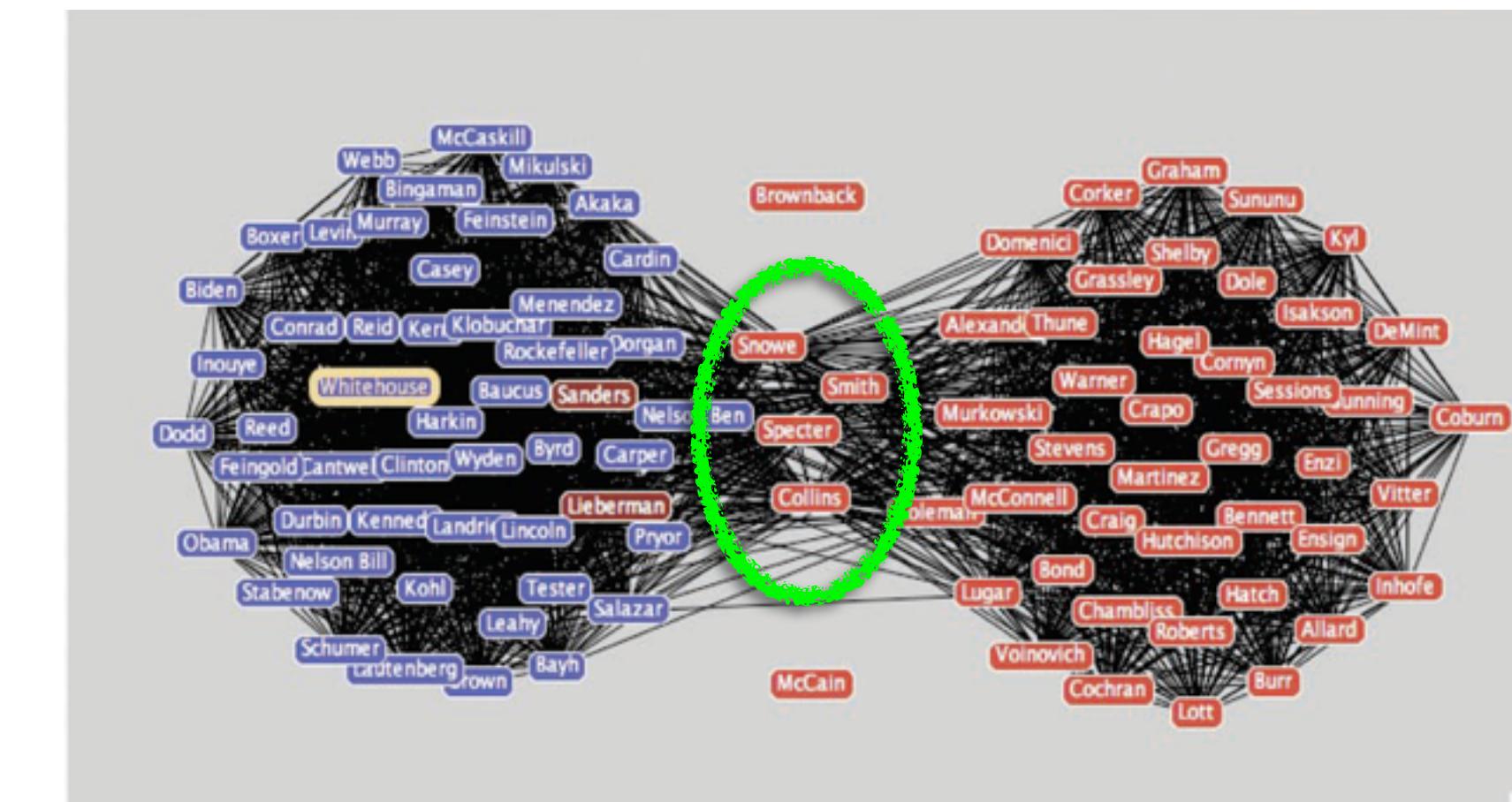
- find clusters
- find high-degree nodes



# Task Abstraction

## Social Network Analysis Domain

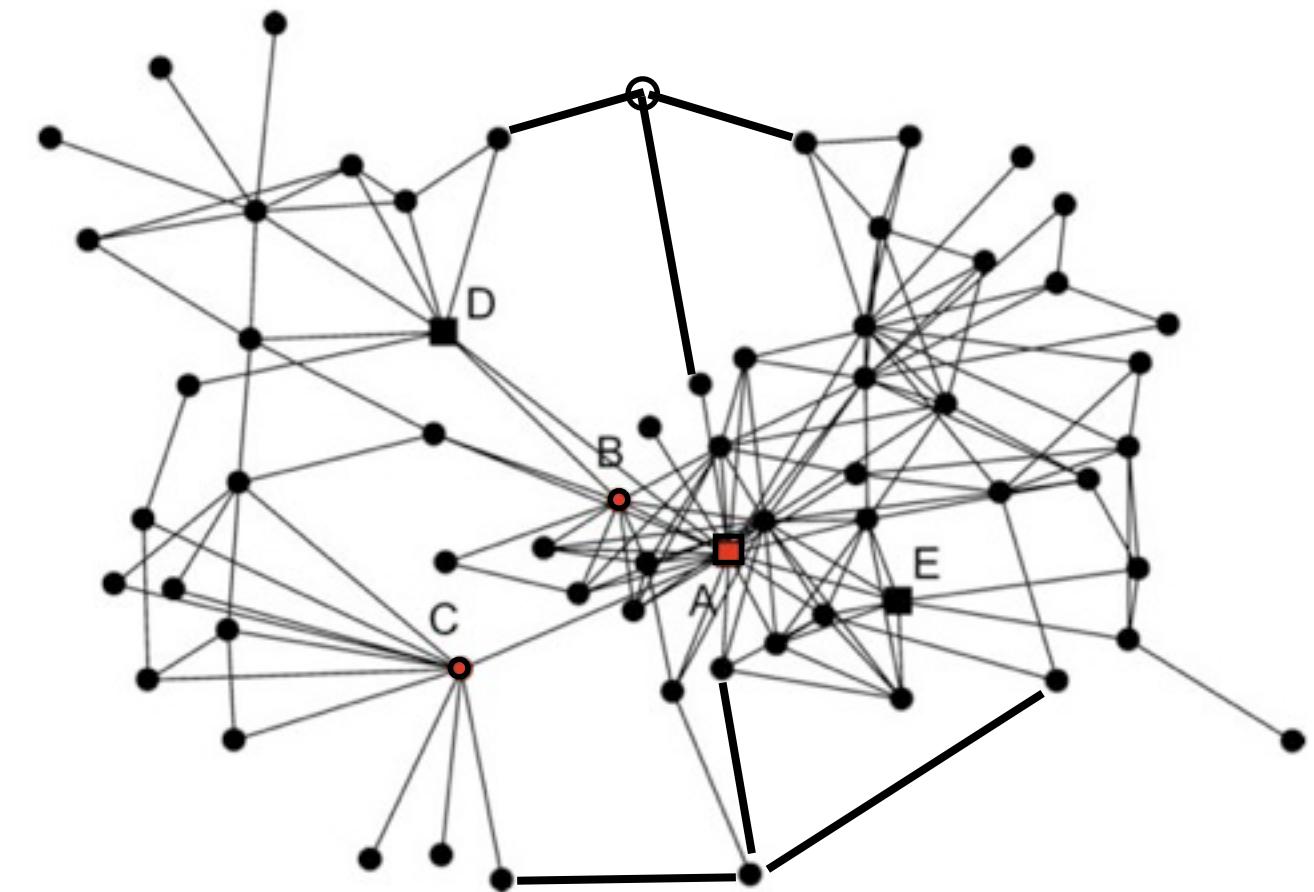
- find clusters
- find high-degree nodes
- find bridge nodes



# Task Abstraction

## Social Network Analysis Domain

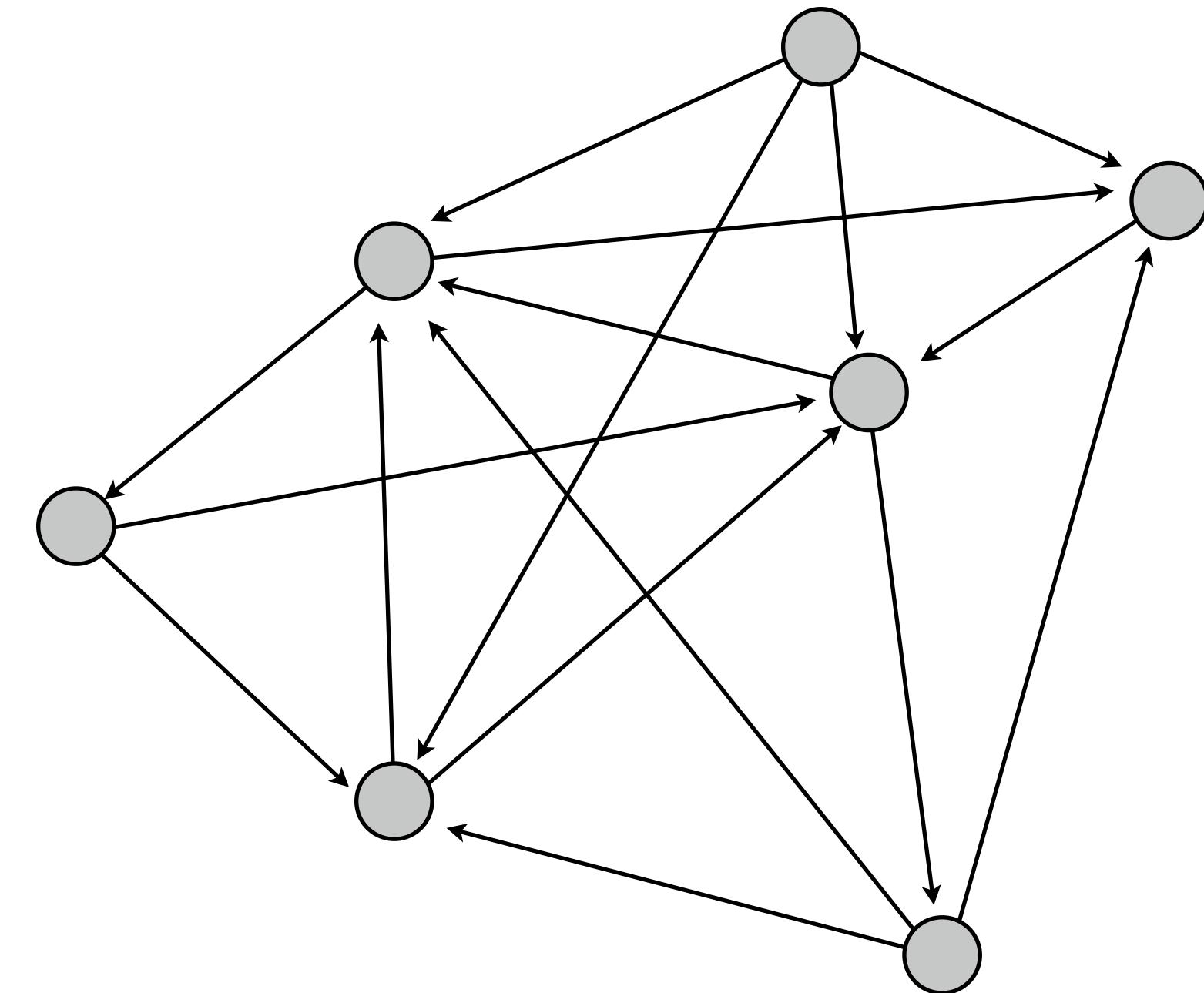
- find clusters
- find high-degree nodes
- find bridge nodes
- understand temporal dynamics
  - passively notice changes



# Data Abstraction

## Social Network Analysis Domain

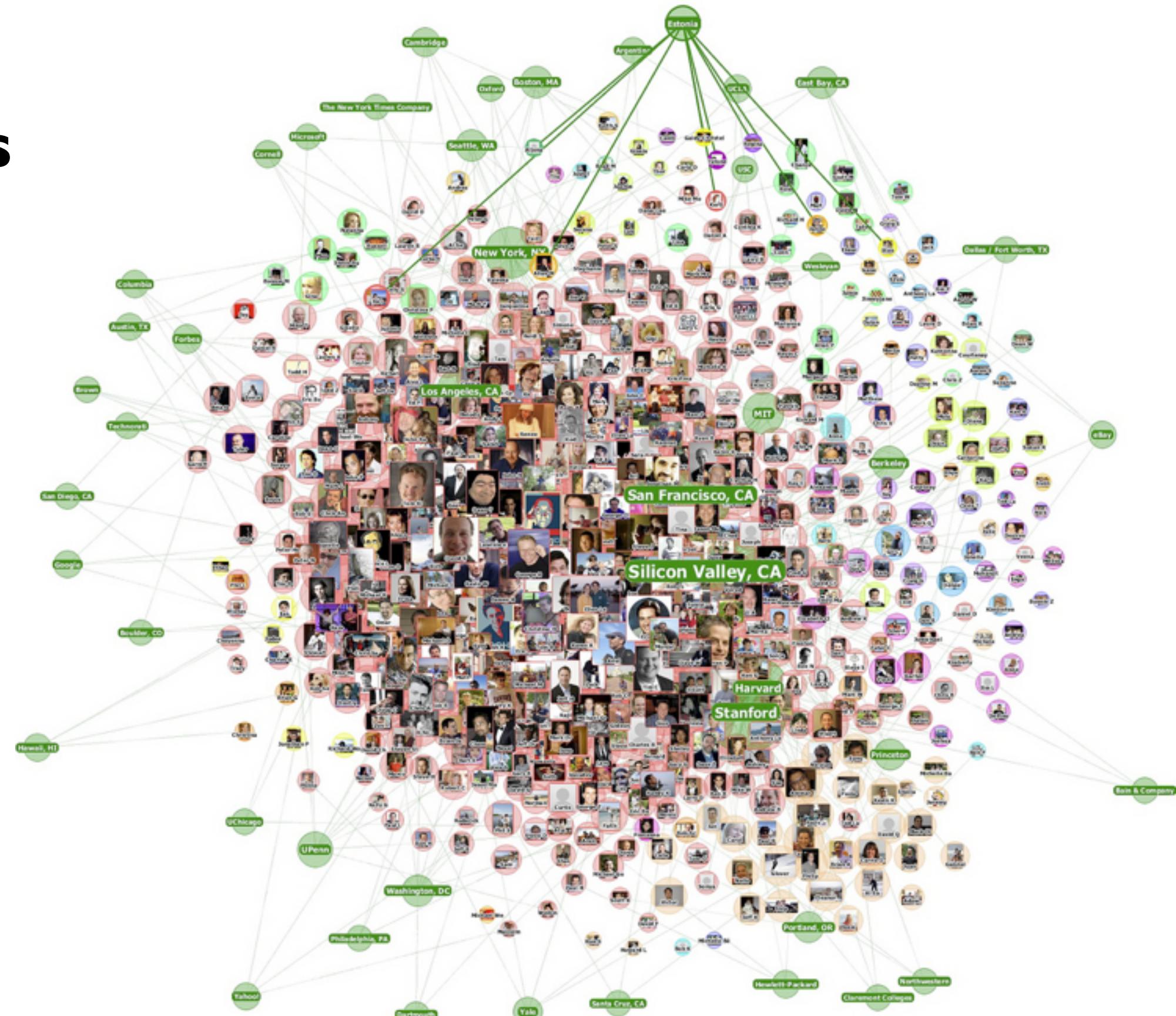
- single graph



# Data Abstraction

# Social Network Analysis

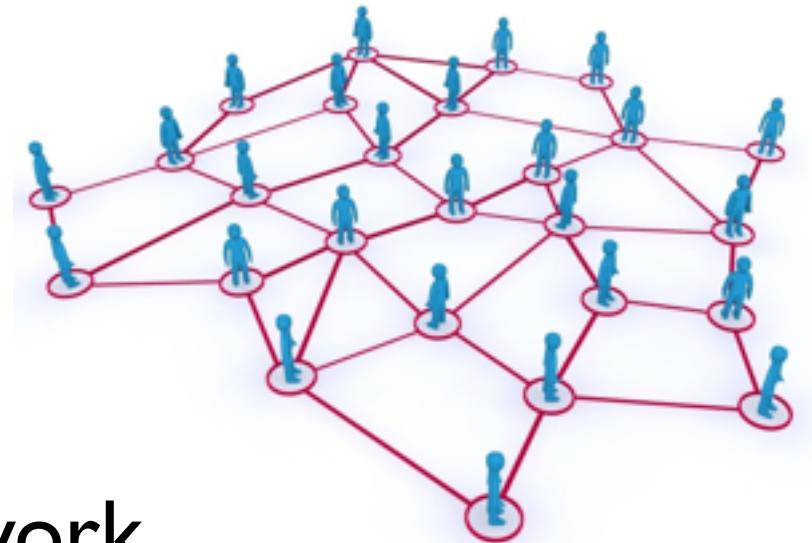
- single graph
  - scalability challenge: nodes



# Abstraction Differences

## Social Network Analysis vs Overlay Network Optimization

- data
  - single network
  - node scalability
    - sparse edges
- task
  - find clusters, high-degree nodes, bridge nodes
  - passive changes



- data
  - three related networks
    - physical, logical, overlay
  - path scalability
    - dense edges, few nodes
- task
  - traffic optimization
  - active changes



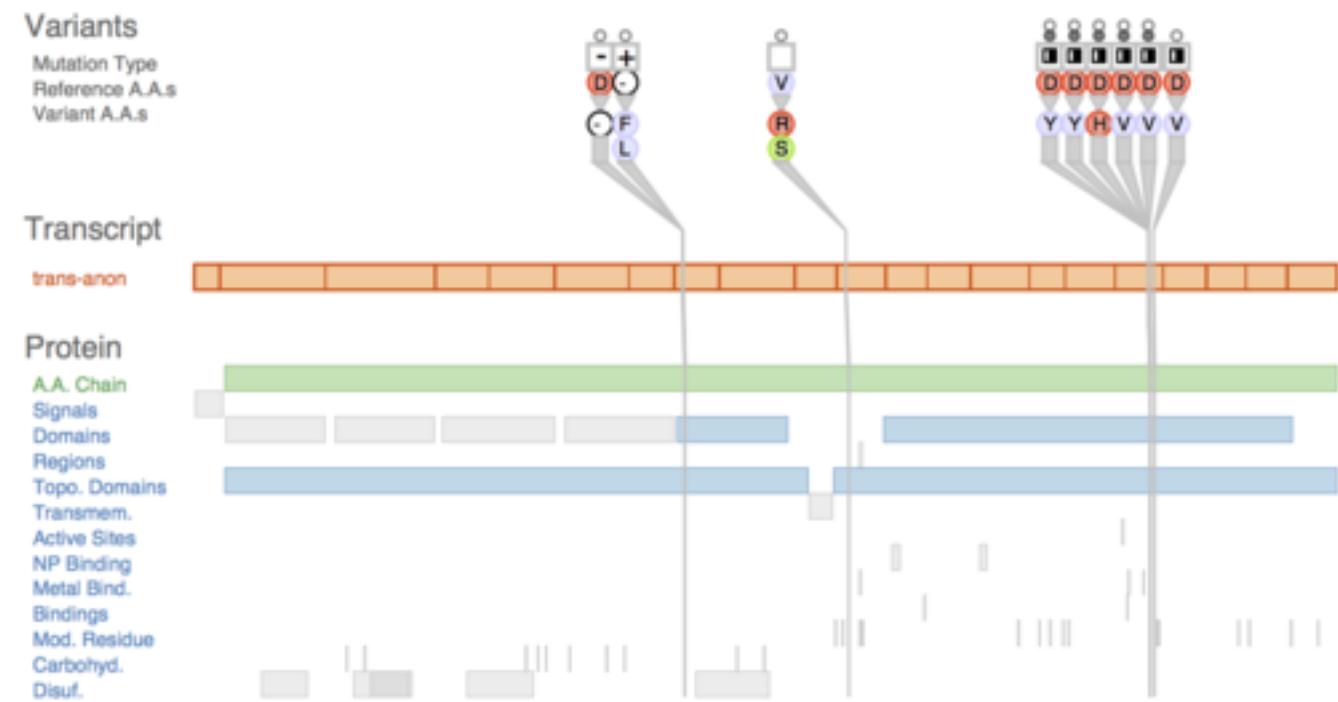
# Variant View

*Visualizing Sequence Variants in their Gene Context*

**joint work with:**

Joel Ferstay, Cydney Nielsen

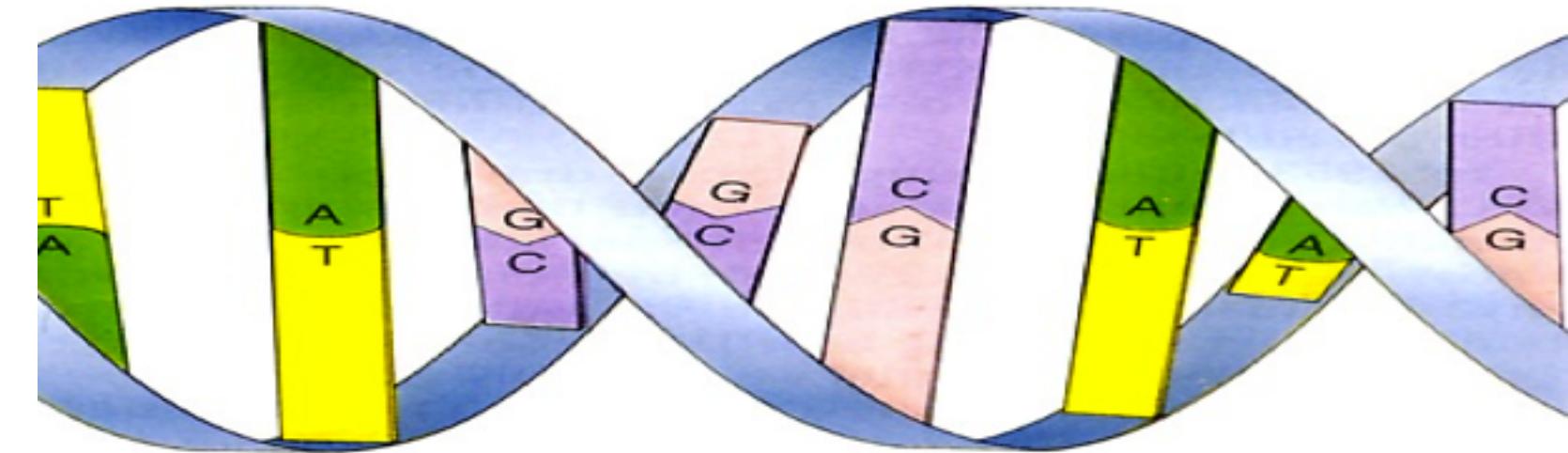
<http://www.cs.ubc.ca/labs/imager/tr/2012/VariantView/>



Variant View: Visualizing Sequence Variants in their Gene Context.  
Ferstay, Nielsen, Munzner. IEEE TVCG 19(12): 2546-2555, 2013 (Proc. InfoVis 2013).

# Sequence Variant Definition

- Sequence variants
  - Difference between reference and given genome



Reference Genome DNA: ATA TGA TCA ACA CTT

Sample 1 Genome DNA: ATA TGG TCA ATA CTT

Harmful?

Sample 2 Genome DNA: ATA TGA TGA ACA CCT

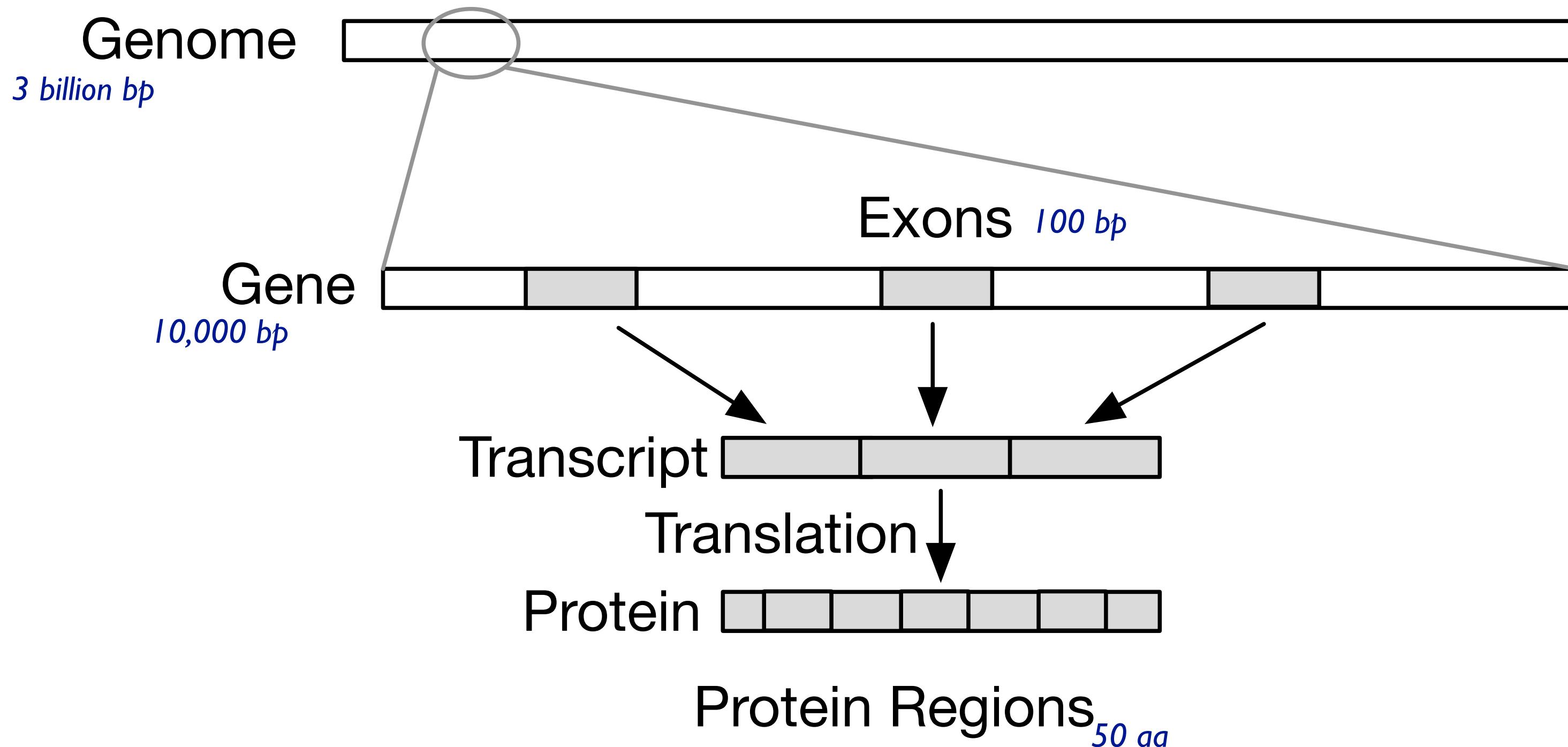
Harmless?

# Cancer Research

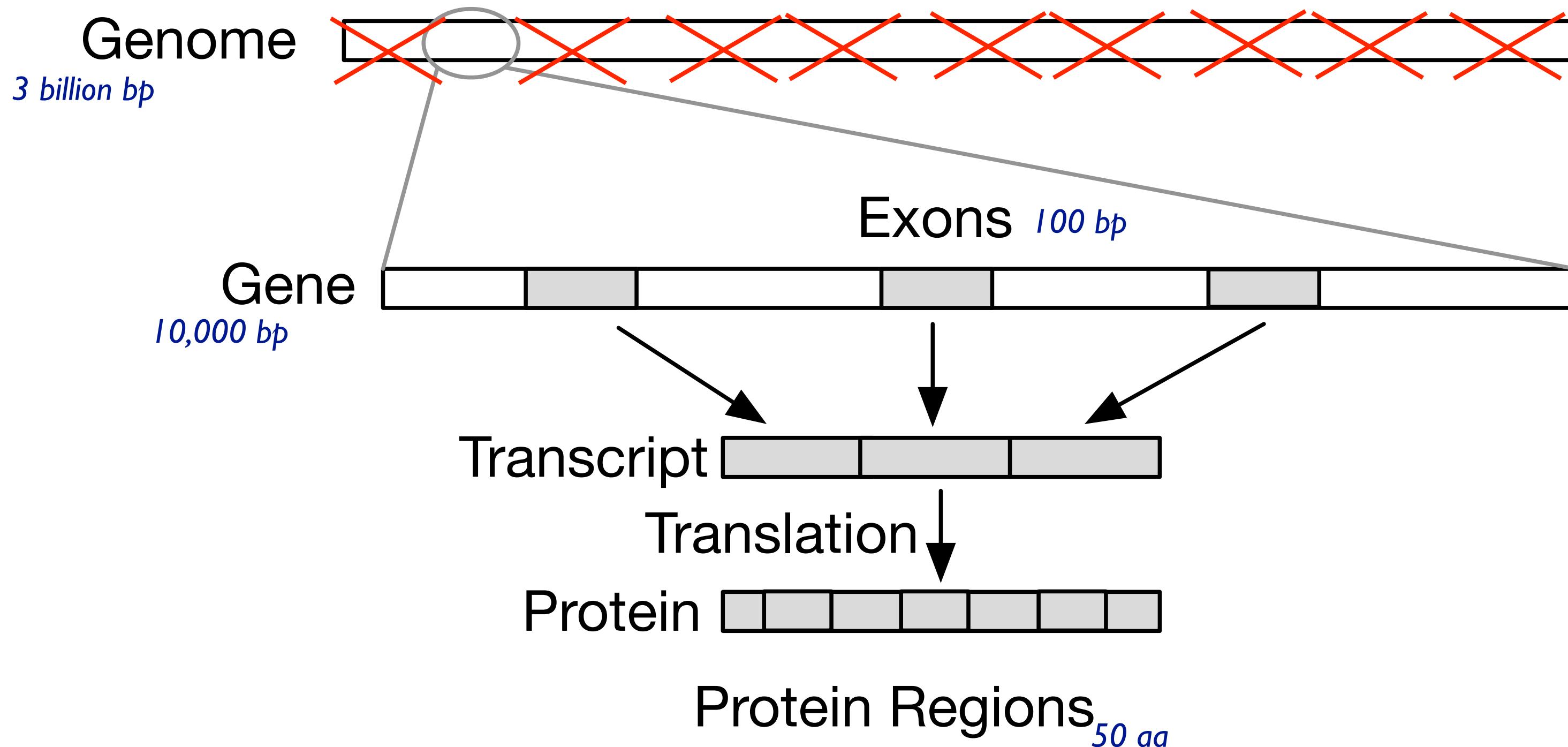
- collaboration with analysts at BC Genome Sciences Center
  - studying genetic basis of leukemia
- driving task
  - discover new candidate genes with harmful variants
- two big questions
  - what to show
    - data abstraction
    - challenge: enormous range of scales in the data
  - how to show it
    - visual encoding idiom

# Abstractions

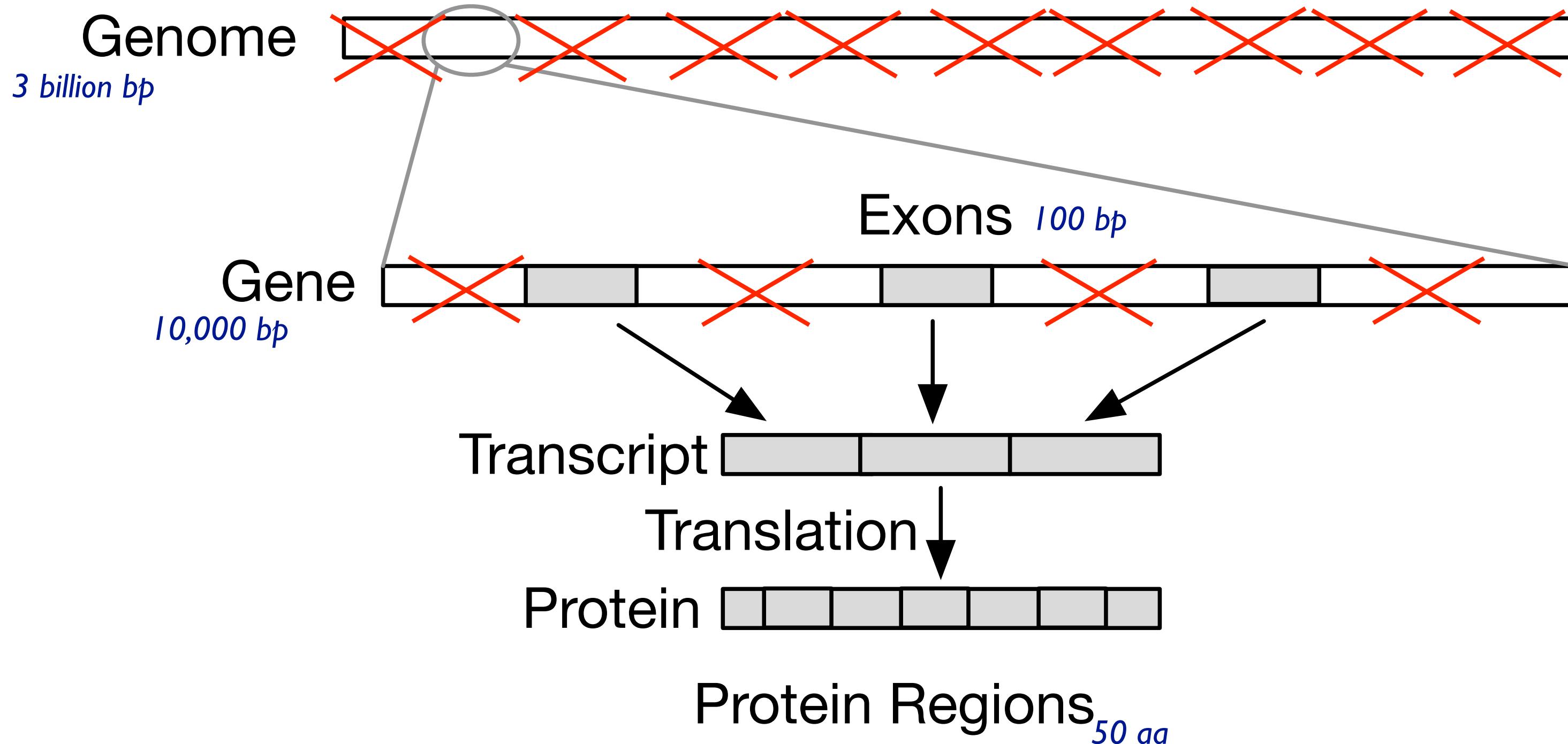
# Data: Filtering to relevant biological levels and scales



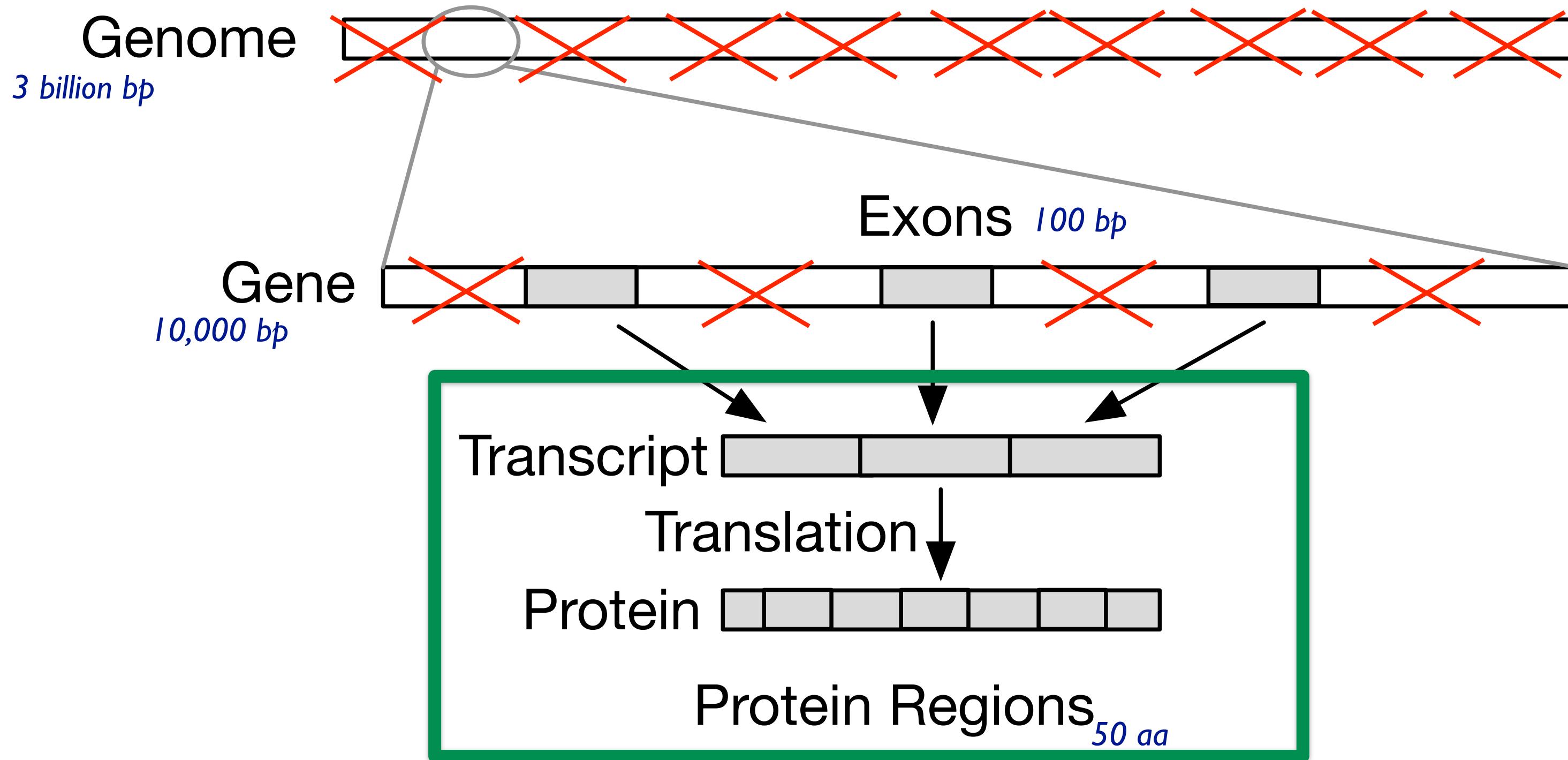
# Filter out whole genome; keep genes



# Filter out non-exon regions

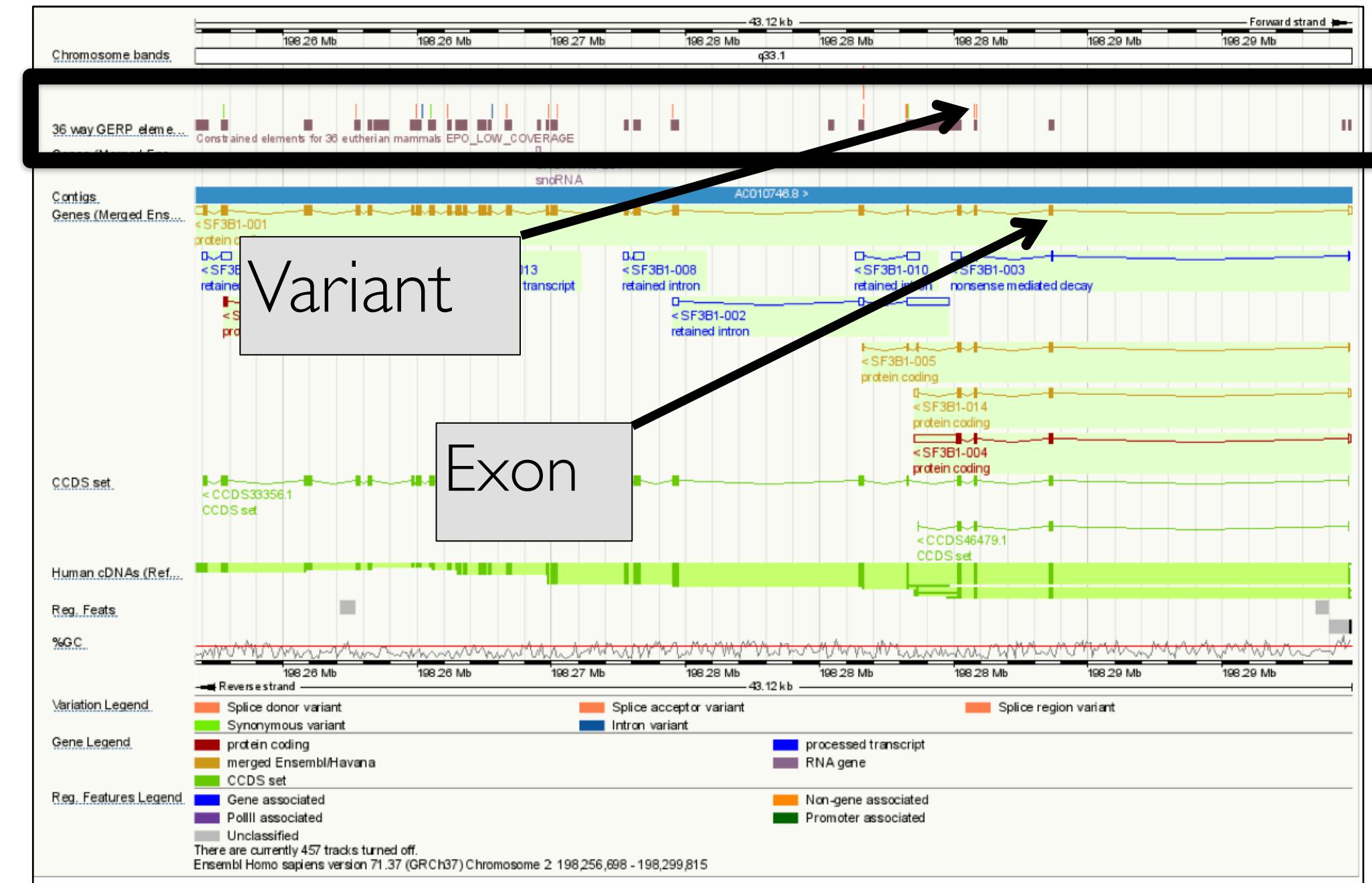


# Data abstraction: highly filtered scope of transcript coordinates



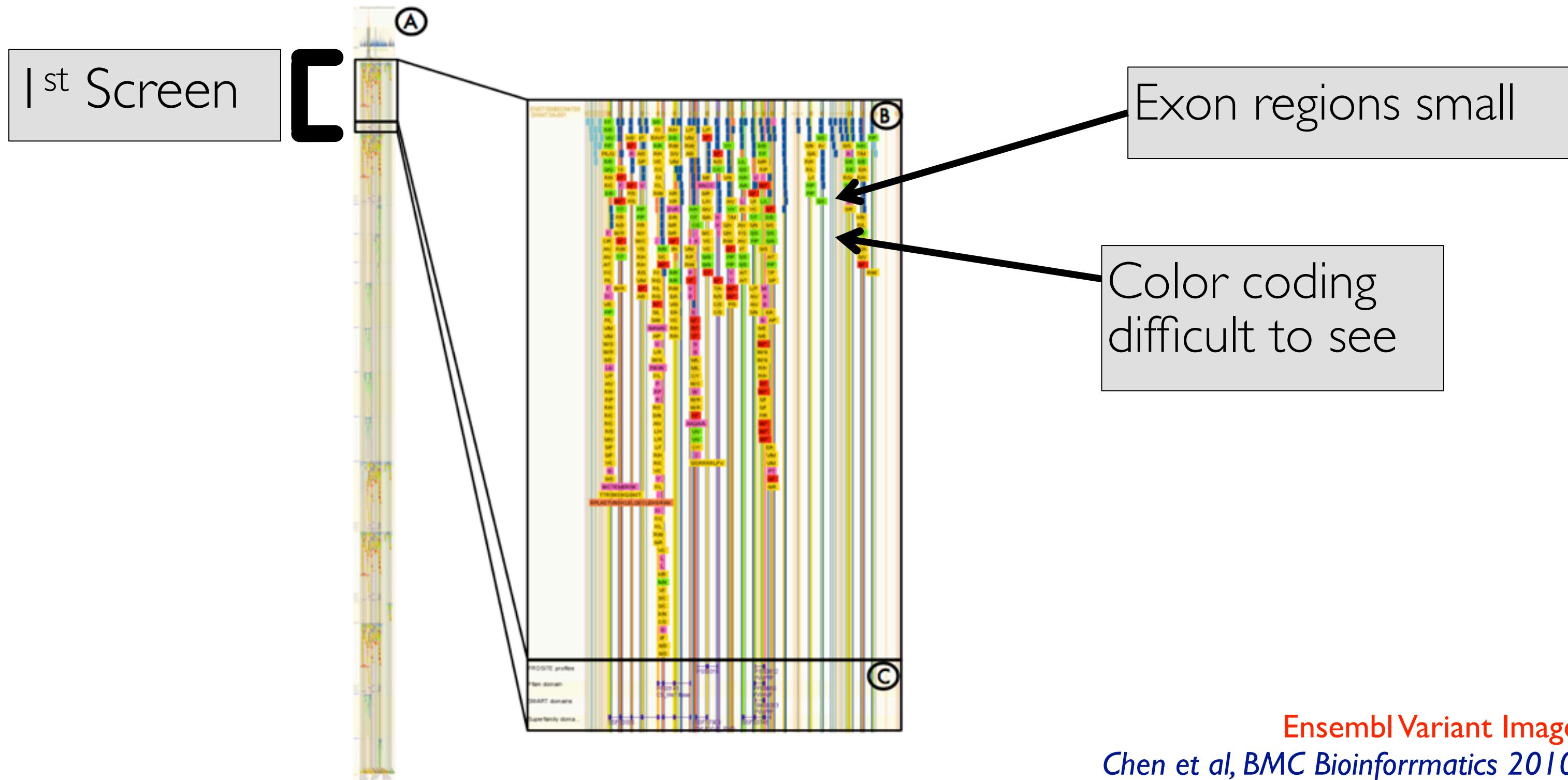
# Dominant paradigm: genome browsers

- strengths: flexible and powerful
  - horizontal tracks: user data
  - shared coordinate system: genome coordinates (bp)
- problems
  - tiny features of interest spread out across large extent
    - must zoom far in to inspect known feature, then zoom out and pan to locate next
    - high cognitive load for interaction
    - must already know where to look



representative example: Ensembl  
Chen et al, BMC Bioinformatics 2010.

# Features of interest small even in variant-specific view



# Idioms

# Variant View

Gene Search:

**A**

Alternative Transcripts: gene-anon (trans-anon)

Variants

Mutation Type  
Reference A.A.s  
Variant A.A.s

Transcript

trans-anon

Protein

A.A. Chain  
Domains  
Regions  
Active Sites  
Bindings  
Mod. Residue

**B**

Variant Data

Patient ID	Chr. Coord.	Ref Base	Var Base	dbSNP129	dbSNP135	dbSNP137	COSMIC	A.A. Chng.	Gene	Ref ID
pid-anon	11288816	G	T	.	.	.	"13028,	G60V	gene-anon	trans-anon
pid-anon	11288816	G	T	.	.	.	"13012,	D61Y	gene-anon	trans-anon
pid-anon	11288819	G	T	.	rs121918	.	13014	A72S	gene-anon	trans-anon
pid-anon	11288819	C	T	.	.	.	"13035,	A72V	gene-anon	trans-anon
pid-anon	11288821	G	C	.	.	.	"13016,	E76Q	gene-anon	trans-anon
pid-anon	11288821	A	G	.	rs121918	.	"13017,	E76G	gene-anon	trans-anon
pid-anon	11288821	G	T	.	.	.	.	E76D	gene-anon	trans-anon
pid-anon	11292688	T	A	.	rs121918	.	"13020,	S502T	gene-anon	trans-anon
pid-anon	11292688	T	G	.	.	.	"13020,	S502A	gene-anon	trans-anon
pid-anon	11292688	C	T	.	.	.	13023	S502L	gene-anon	trans-anon

**C**

Sort By Gene:

Alpha Cluster Score Variant Count

DNMT3A (NM_022552)
IDH2 (NM_002168)
FLT3 (NM_004119)
ANKRD36 (NM_001164315)
ARID1B (NM_017519)
STAG2 (NM_001042749)
TNRC18 (NM_001080495)
WT1 (NM_000378)
ABCA13 (NM_152701)
CEBPA (NM_004364)
TET2 (NM_001127208)
DNAH10 (NM_207437)
GPSM1 (NM_015597)
ASXL1 (NM_015338)
DNAH1 (NM_015512)
DNAH6 (NM_001370)
FAT1 (NM_005245)
MDN1 (NM_014611)
PTPN11 (NM_002834)
SYNE1 (NM_033071)
ALMS1 (NM_015120)
C10orf68 (NM_024688)
CCDC88C (NM_001080414)
DNAH11 (NM_003777)
DNAH3 (NM_017539)
DNAH9 (NM_001372)

# Variant View

Information-dense single gene view

Gene Search:  Submit

**A**

Alternative Transcripts: gene-anon (trans-anon)

Variants

Mutation Type  
Reference A.A.s  
Variant A.A.s

Transcript

trans-anon

Protein

A.A. Chain  
Domains  
Regions  
Active Sites  
Bindings  
Mod. Residue

Variant Data

Patient ID	Chr. Coord.	Ref Base	Var Base	dbSNP129	dbSNP135	dbSNP137	COSMIC	A.A. Chng.	Gene	Ref ID
pid-anon	11288816	G	T	.	.	.	"13028,	G60V	gene-anon	trans-anon
pid-anon	11288816	G	T	.	.	.	"13012,	D61Y	gene-anon	trans-anon
pid-anon	11288819	G	T	.	rs121918	.	13014	A72S	gene-anon	trans-anon
pid-anon	11288819	C	T	.	.	.	"13035,	A72V	gene-anon	trans-anon
pid-anon	11288821	G	C	.	.	.	"13016,	E76Q	gene-anon	trans-anon
pid-anon	11288821	A	G	.	rs121918	.	"13017,	E76G	gene-anon	trans-anon
pid-anon	11288821	G	T	.	.	.	.	E76D	gene-anon	trans-anon
pid-anon	11292688	T	A	.	rs121918	.	"13020,	S502T	gene-anon	trans-anon
pid-anon	11292688	T	G	.	.	.	"13020,	S502A	gene-anon	trans-anon
pid-anon	11292688	C	T	.	.	.	13023	S502L	gene-anon	trans-anon

**B**

**C**

Sort By Gene:

Alpha Cluster Score Variant Count

- DNMT3A (NM\_022552)
- IDH2 (NM\_002168)
- FLT3 (NM\_004119)
- ANKRD36 (NM\_001164315)
- ARID1B (NM\_017519)
- STAG2 (NM\_001042749)
- TNRC18 (NM\_001080495)
- WT1 (NM\_000378)
- ABCA13 (NM\_152701)
- CEBPA (NM\_004364)
- TET2 (NM\_001127208)
- DNAH10 (NM\_207437)
- GPSM1 (NM\_015597)
- ASXL1 (NM\_015338)
- DNAH1 (NM\_015512)
- DNAH6 (NM\_001370)
- FAT1 (NM\_005245)
- MDN1 (NM\_014611)
- PTPN11 (NM\_002834)
- SYNE1 (NM\_033071)
- ALMS1 (NM\_015120)
- C10orf68 (NM\_024688)
- CCDC88C (NM\_001080414)
- DNAH11 (NM\_003777)
- DNAH3 (NM\_017539)
- DNAH9 (NM\_001372)

6 |

# Variant View

Information-dense single gene view

**A**

Gene Search:  Submit

Alternative Transcripts: gene-anon (trans-anon)

Variants

Mutation Type  
Reference A.A.s  
Variant A.A.s

Transcript

trans-anon

Protein

A.A. Chain  
Domains  
Regions  
Active Sites  
Bindings  
Mod. Residue

Variant Data

Patient ID	Chr. Coord.	Ref Base	Var Base	dbSNP129	dbSNP135	dbSNP137	COSMIC	A.A. Ch.	Gene	Protein
pid-anon	11288816	G	T	.	.	.	"13028,	G60V	gene-anon	trans-anon
pid-anon	11288816	G	T	.	.	.	"13012,	D61Y	gene-anon	trans-anon
pid-anon	11288819	G	T	.	rs121918	.	13014	A72S	gene-anon	trans-anon
pid-anon	11288819	C	T	.	.	.	"13035,	A72V	gene-anon	trans-anon
pid-anon	11288821	G	C	.	.	.	"13016,	E76Q	gene-anon	trans-anon
pid-anon	11288821	A	G	.	rs121918	.	13017,	E76G	gene-anon	trans-anon
pid-anon	11288821	G	T	.	.	.	.	E76D	gene-anon	trans-anon
pid-anon	11292688	T	A	.	rs121918	.	"13020,	S502T	gene-anon	trans-anon
pid-anon	11292688	T	G	.	.	.	"13020,	S502A	gene-anon	trans-anon
pid-anon	11292688	C	T	.	.	.	13023	S502L	gene-anon	trans-anon

**C**

Sort By Gene:

Alpha Cluster Score Variant Count

- DNMT3A (NM\_022552)
- IDH2 (NM\_002168)
- FLT3 (NM\_004119)
- ANKRD36 (NM\_001164315)
- ARID1B (NM\_017519)
- STAG2 (NM\_001042749)
- TNRC18 (NM\_001080495)
- WT1 (NM\_000378)
- ABCA13 (NM\_152701)
- CEBPA (NM\_004364)
- TET2 (NM\_001127208)
- DNAH10 (NM\_207437)
- GPSM1 (NM\_015597)
- ASXL1 (NM\_015338)
- DNAH1 (NM\_015512)
- DNAH6 (NM\_001370)

No need for pan and zoom

**62**

# Variant View

Sorting metrics guide gene navigation

The Variant View interface displays gene navigation tools and variant data.

**Sorting Metrics:** The top right corner shows a dropdown menu for sorting genes by "Cluster Score" (selected), "Alpha", or "Variant Count".

**Alternative Transcripts:** Shows gene-anon (trans-anon) transcripts with variants highlighted.

**Variants:** Details mutation type, reference and variant amino acids, and protein domains.

**Transcript:** Shows the trans-anon transcript structure.

**Protein:** Shows the protein chain with domains and active sites.

**Variant Data:** A table listing patient ID, chromosomal coordinates, reference and variant bases, dbSNP IDs, COSMIC ID, amino acid change, gene name, and reference ID for multiple variants.

**Gene List:** A vertical list of genes sorted by cluster score, including DNMT3A, IDH2, FLT3, ANKRD36, ARID1B, STAG2, TNRC18, WT1, ABCA13, CEBPA, TET2, DNAH10, GPSM1, ASXL1, DNAH1, DNAH6, FAT1, MDN1, PTPN11, SYNE1, ALMS1, C10orf68, CCDC88C, DNAH11, DNAH3, and DNAH9.

Patient ID	Chr. Coord.	Ref Base	Var Base	dbSNP129	dbSNP135	dbSNP137	COSMIC	A.A. Chng.	Gene	Ref ID
pid-anon	11288816	G	T	.	.	.	"13028,	G60V	gene-anon	trans-anon
pid-anon	11288816	G	T	.	.	.	"13012,	D61Y	gene-anon	trans-anon
pid-anon	11288819	G	T	.	rs121918	.	13014	A72S	gene-anon	trans-anon
pid-anon	11288819	C	T	.	.	.	"13035,	A72V	gene-anon	trans-anon
pid-anon	11288821	G	C	.	.	.	"13016,	E76Q	gene-anon	trans-anon
pid-anon	11288821	A	G	.	rs121918	.	"13017,	E76G	gene-anon	trans-anon
pid-anon	11288821	G	T	.	.	.	.	E76D	gene-anon	trans-anon
pid-anon	11292688	T	A	.	rs121918	.	"13020,	S502T	gene-anon	trans-anon
pid-anon	11292688	T	G	.	.	.	"13020,	S502A	gene-anon	trans-anon
pid-anon	11292688	C	T	.	.	.	13023	S502L	gene-anon	trans-anon

# Variant View

Sorting metrics guide gene navigation

The screenshot illustrates the Variant View interface, which integrates gene navigation and variant filtering.

**Panel A:** On the right, a sidebar lists genes sorted by a metric, with "Cluster Score" as the active filter. An arrow labeled "A" points from the "Variant Count" header to this list.

**Panel B:** In the bottom left, a table displays variants from dbSNP135, dbSNP137, COSMIC, and other sources. An arrow labeled "B" points from the "Variant Data" header to this table.

**Panel C:** On the right, a detailed view of the gene *DNMT3A* shows alternative transcripts (gene-anon and trans-anon), variants with their mutation types and amino acid changes, and the resulting protein sequence. An arrow labeled "C" points from the gene name to this panel.

**Panel D:** At the bottom left, a large text box states "Control what shows up here".

**Variant Data Table:**

Source	rsID	Chromosome	Position	Ref	Alt	dbSNP135	dbSNP137	COSMIC	A.A. Chng.	Gene	Ref ID
pid-anon	11288819	C	T	-	-	"13028,	"13012,	"13014	G60V	gene-anon	trans-anon
pid-anon	11288821	G	C	-	-	"13035,	"13016,	"13017,	D61Y	gene-anon	trans-anon
pid-anon	11288821	A	G	-	-	"13016,	E76Q	E76G	A72S	gene-anon	trans-anon
pid-anon	11288821	G	T	-	-	"13035,	E76Q	E76D	A72V	gene-anon	trans-anon
pid-anon	11292688	T	A	-	-	"13020,	S502T	S502A	E76D	gene-anon	trans-anon
pid-anon	11292688	T	G	-	-	"13020,	S502T	S502L	S502A	gene-anon	trans-anon
pid-anon	11292688	C	T	-	-	"13023	S502L	S502L	S502L	gene-anon	trans-anon

# Variant View

Gene Search:

Alternative Transcripts: gene-anon (trans-anon)

Variants

Mutation Type  
Reference A.A.s  
Variant A.A.s

Transcript

trans-anon

Protein

A.A. Chain  
Domains  
Regions  
Active Sites  
Bindings  
Mod. Residue

Variant Data

Patient ID	Chr. Coord.	Ref Base	Var Base	dbSNP129	dbSNP135	dbSNP137	COSMIC	A.A. Chng.	Gene	RefSeq ID
pid-anon	11288816	G	T	.	.	.	"13028,	G60V	gene-anon	trans-anon
pid-anon	11288816	G	T	.	.	.	"13012,	D61Y		
pid-anon	11288819	G	T	.	rs121918	.	"13014	A72S		
pid-anon	11288819	C	T	.	.	.	"13035,	A72T		
pid-anon	11288821	G	C	.	.	.	"13016,	E76Q		
pid-anon	11288821	A	G	.	rs121918	.	"13017,	E76G		
pid-anon	11288821	G	T	.	.	.	E76D		gene-anon	trans-anon
pid-anon	11292688	T	A	.	rs121918	.	"13020,	S502T	gene-anon	trans-anon
pid-anon	11292688	T	G	.	.	.	"13020,	S502A	gene-anon	trans-anon
pid-anon	11292688	C	T	.	.	.	13023	S502L	gene-anon	trans-anon

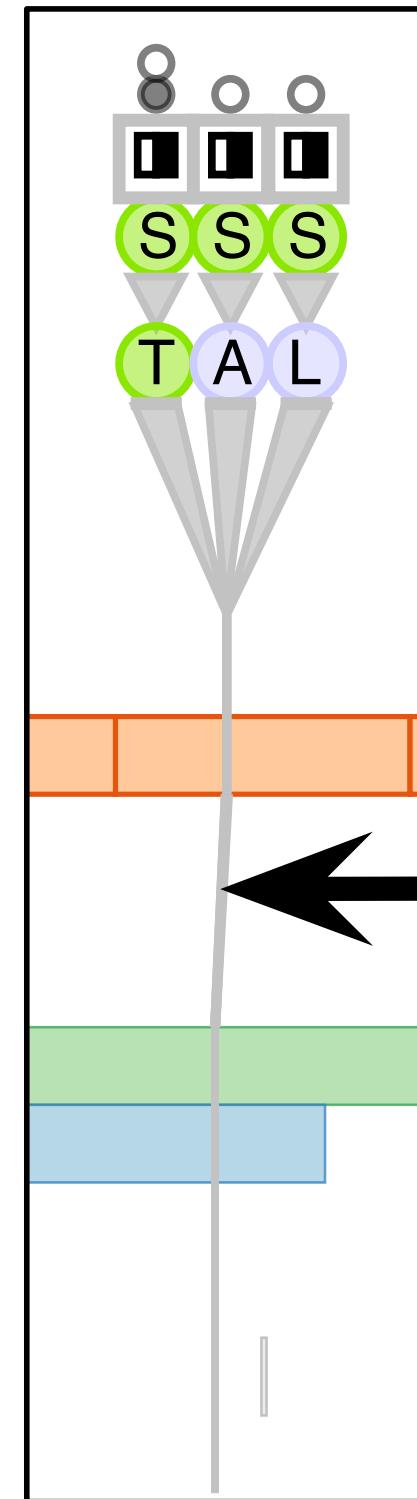
Sort By Gene:  
 Alpha  Cluster Score  Variant Count

(A) (B) (C)

Peripheral supporting data

# Design information-dense visual encoding

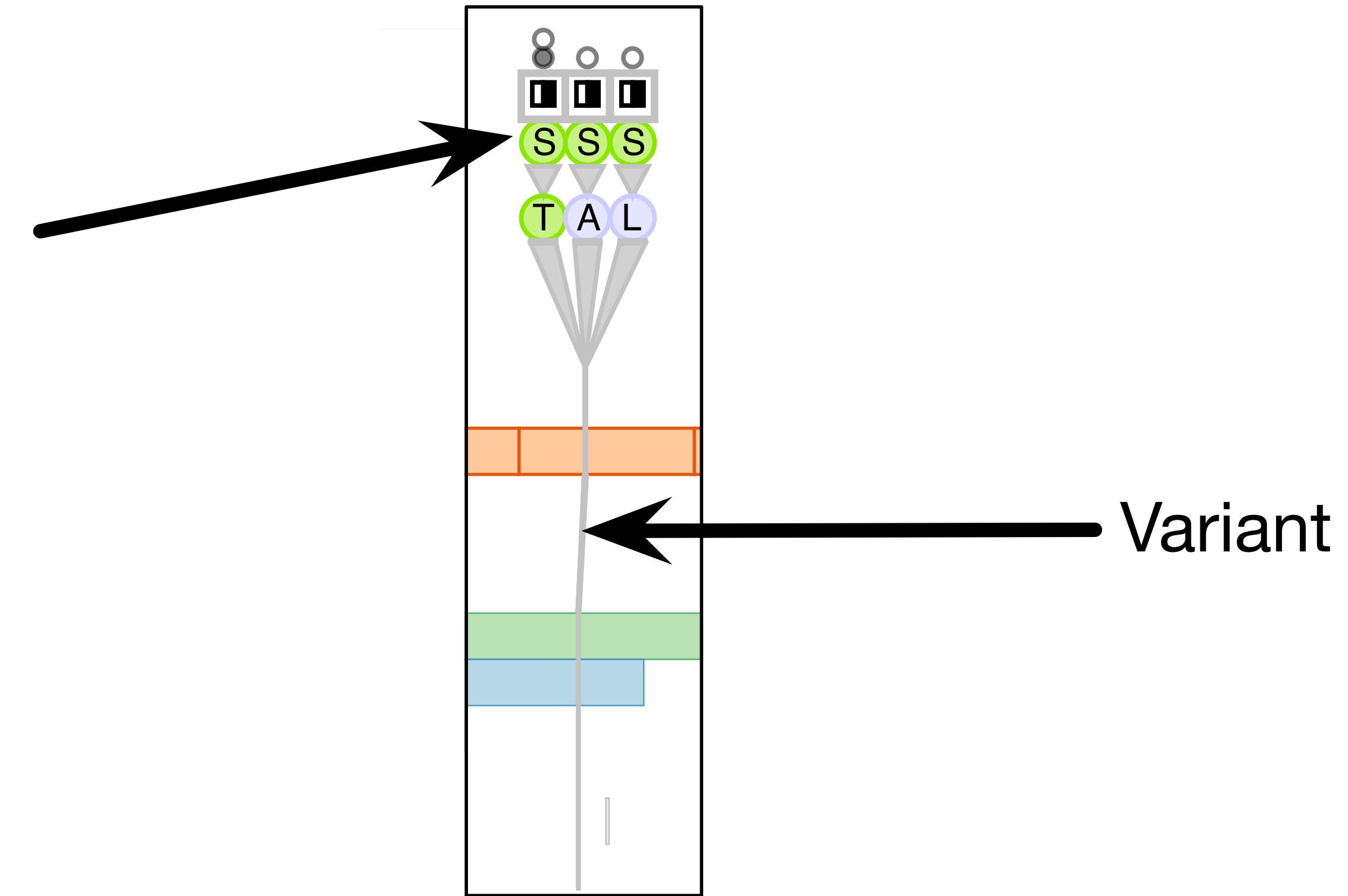
- show all attributes necessary for variant analysis
  - match salience with importance for analysis task
- variant not just a thin line!
- emphasize with high salience
  - collocated variants fan out at top
  - grey variant vertical stroke intersects horizontal colored protein regions



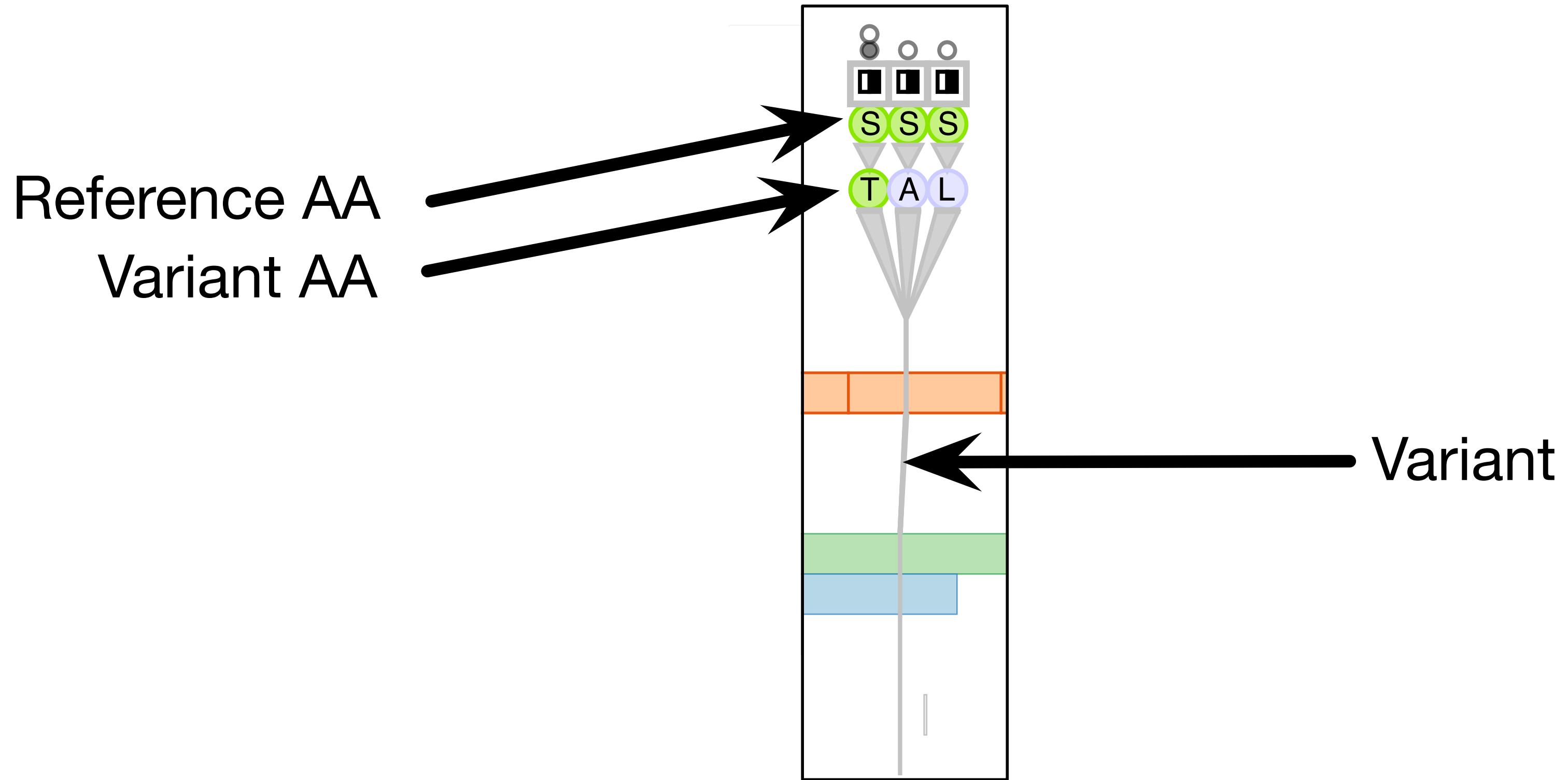
Variant

# Design information-dense visual encoding

Reference AA



# Design information-dense visual encoding



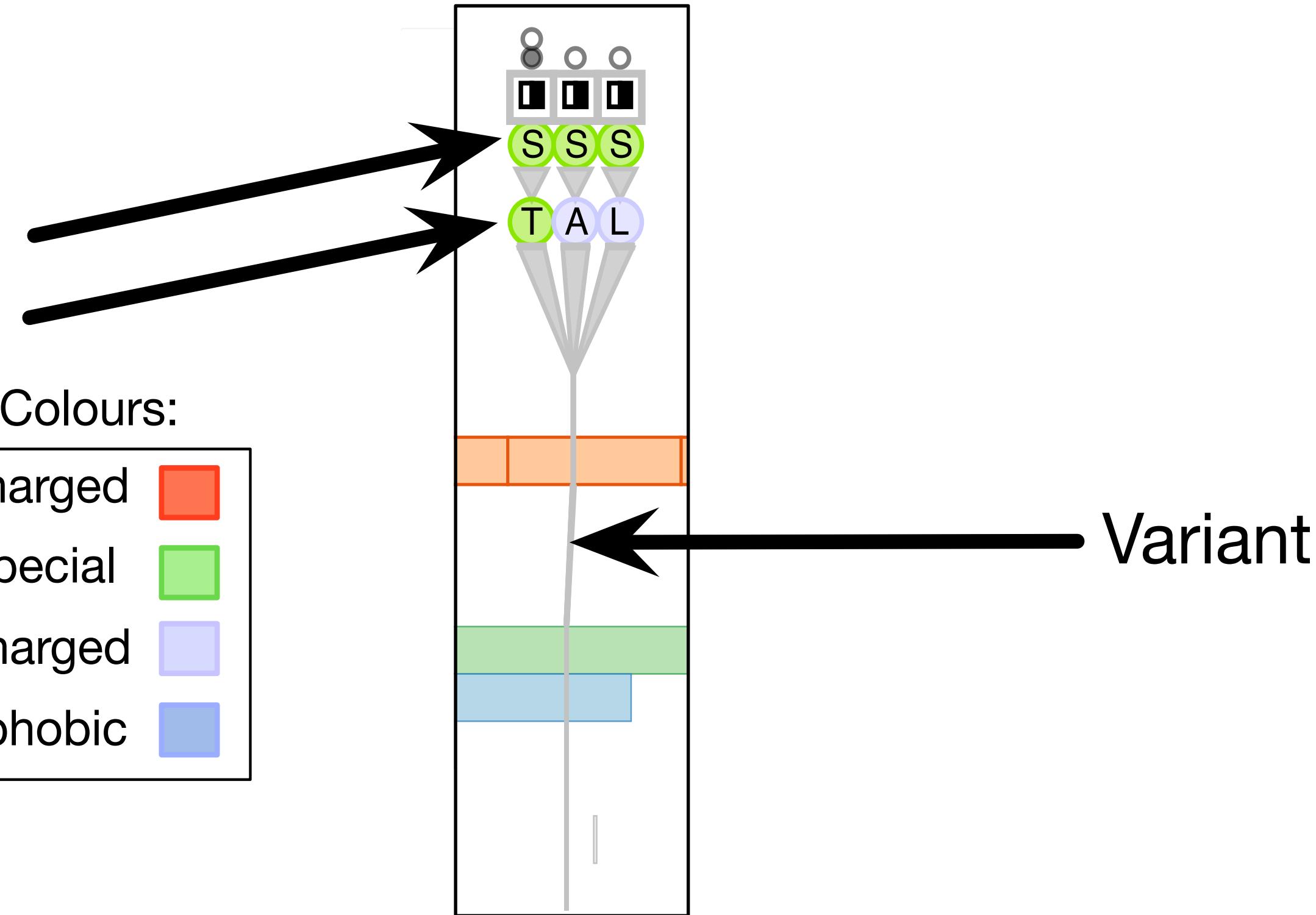
# Design information-dense visual encoding

Reference AA

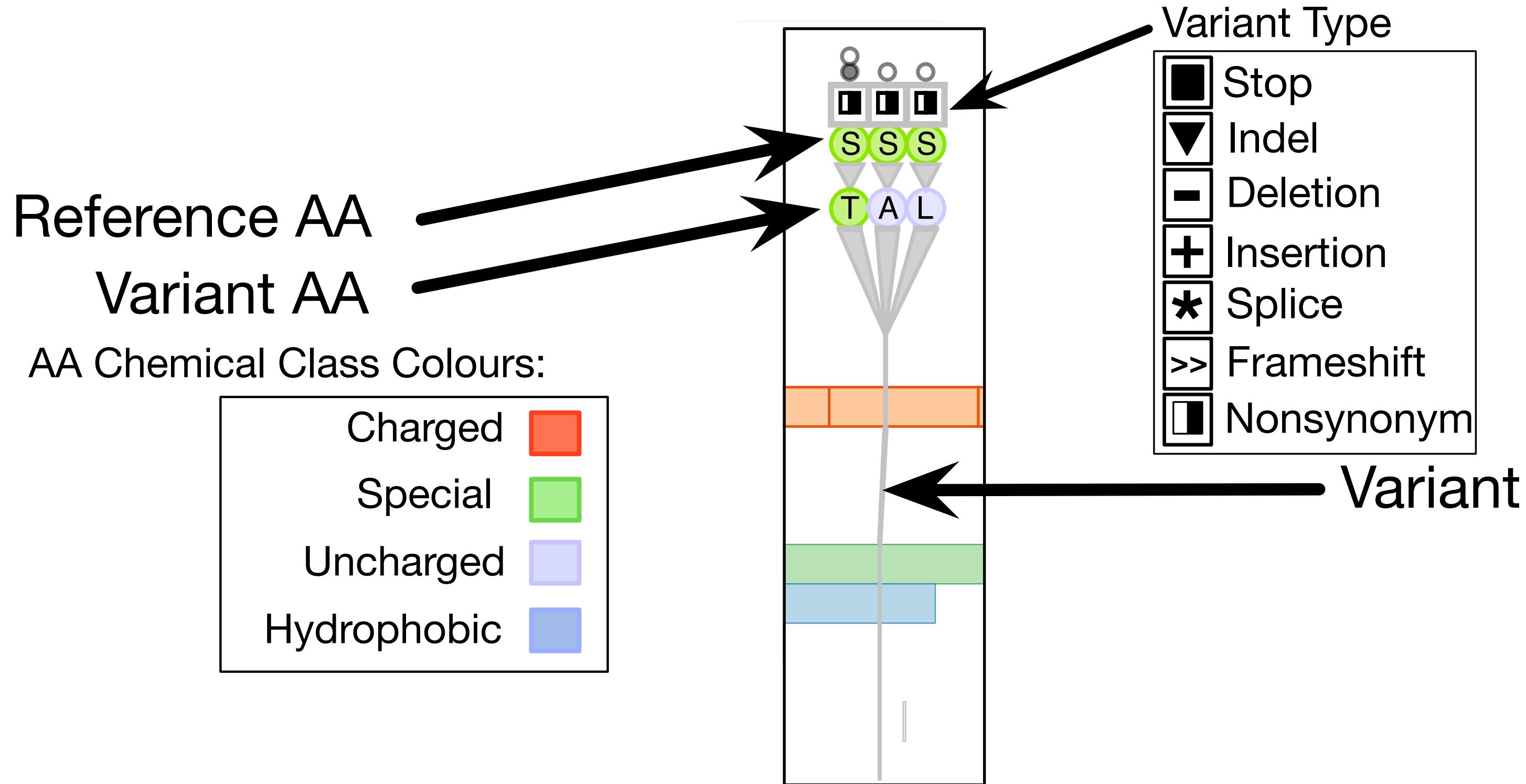
Variant AA

AA Chemical Class Colours:

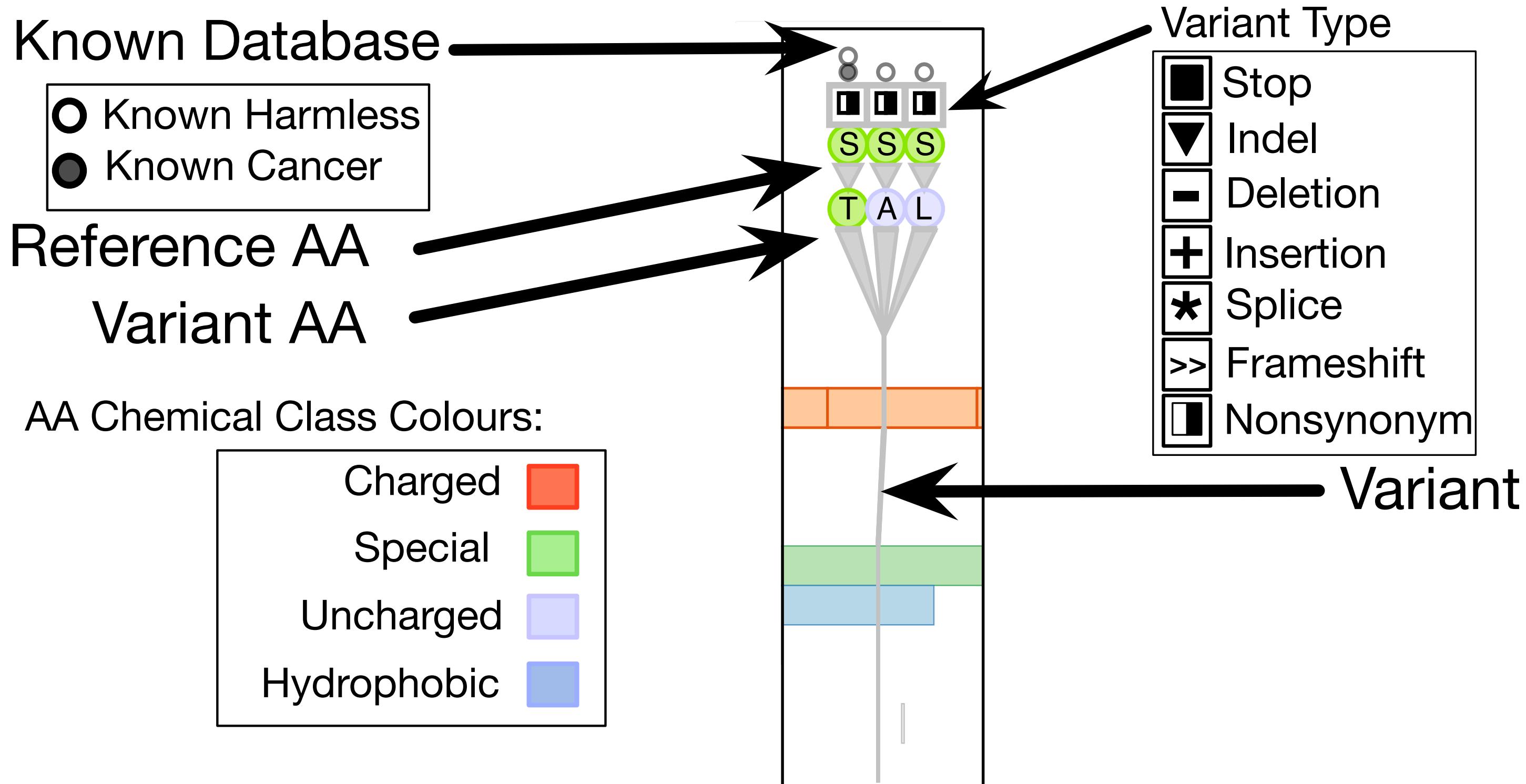
Charged	
Special	
Uncharged	
Hydrophobic	



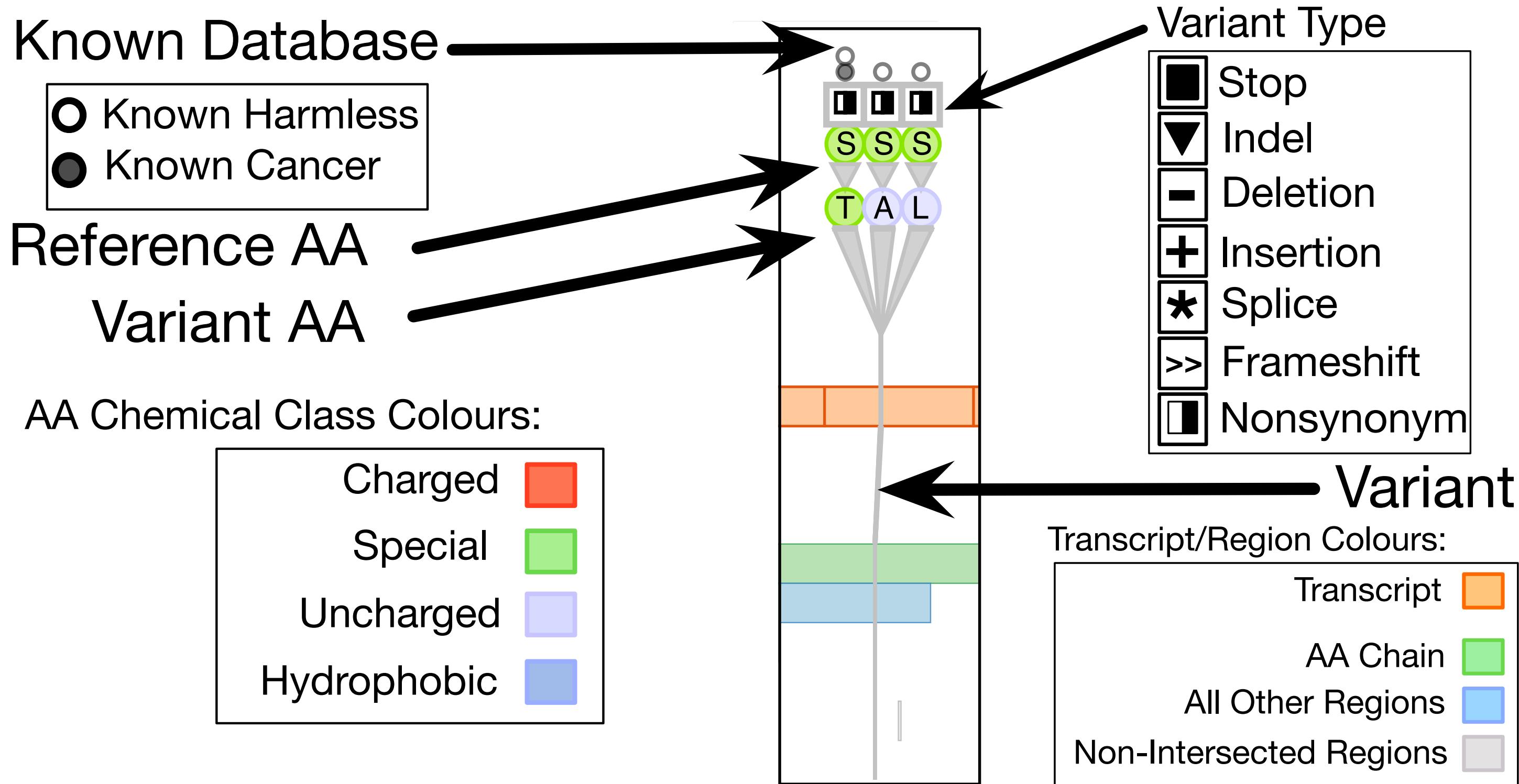
# Design information-dense visual encoding



# Design information-dense visual encoding

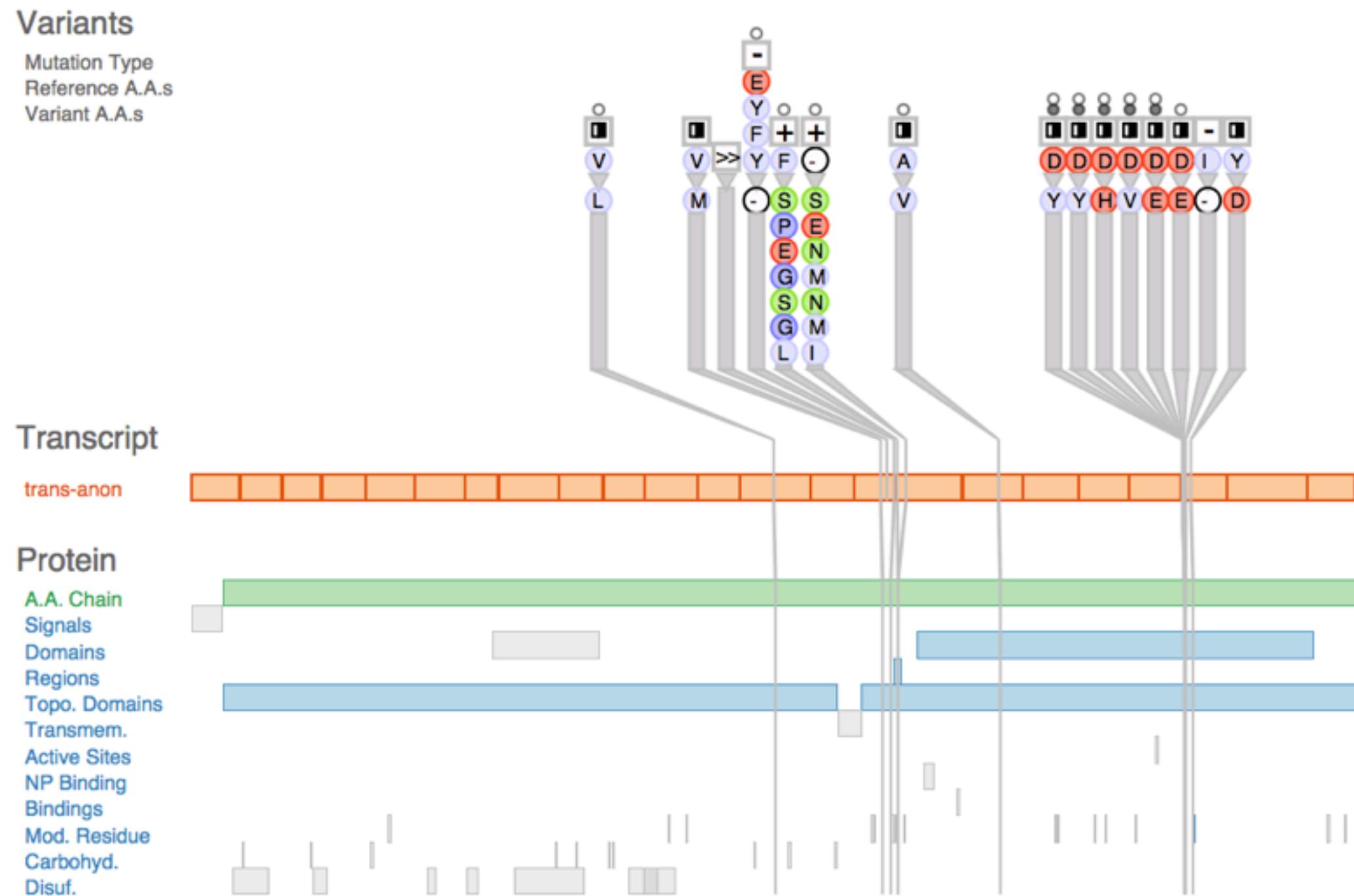


# Design information-dense visual encoding

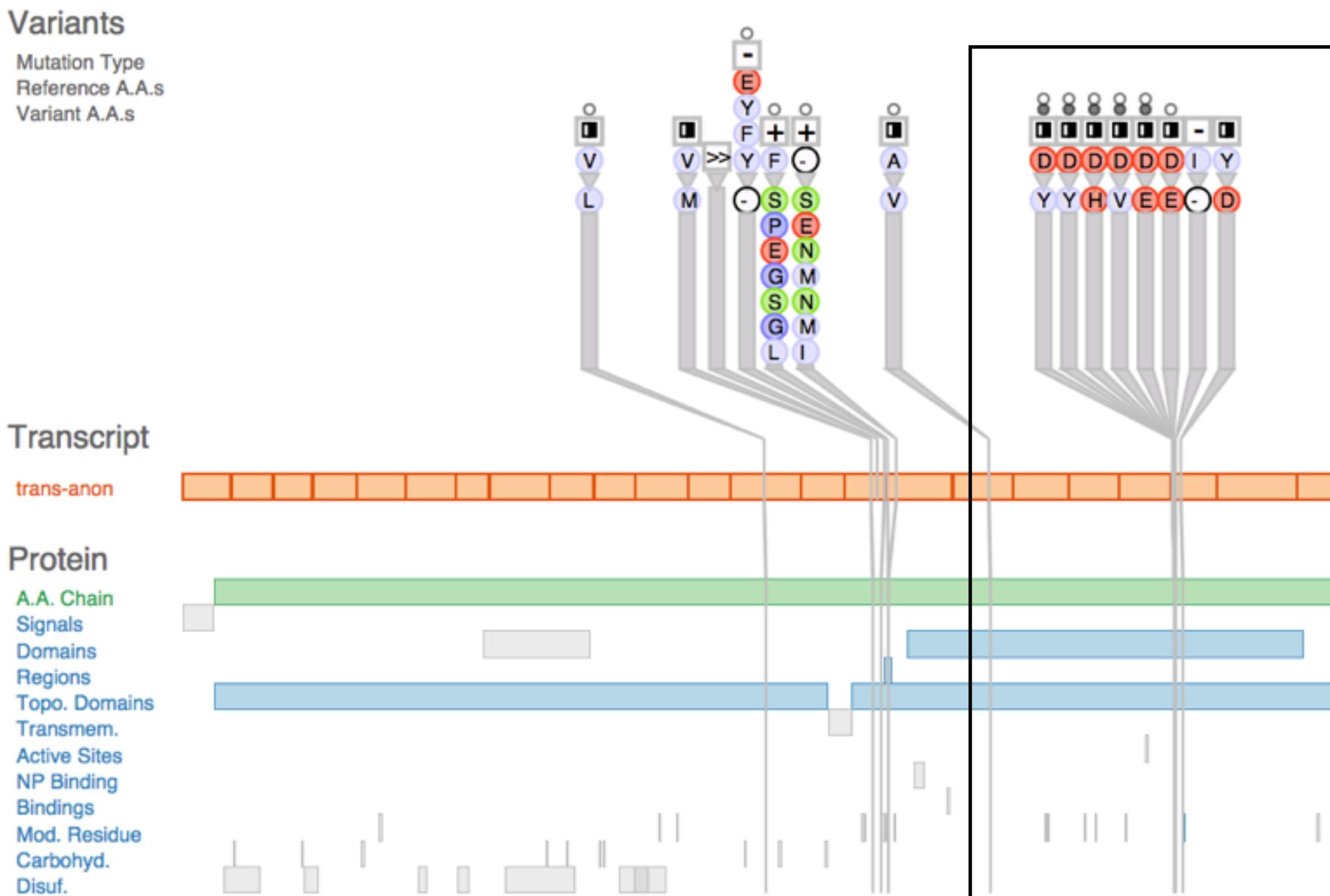


# Results

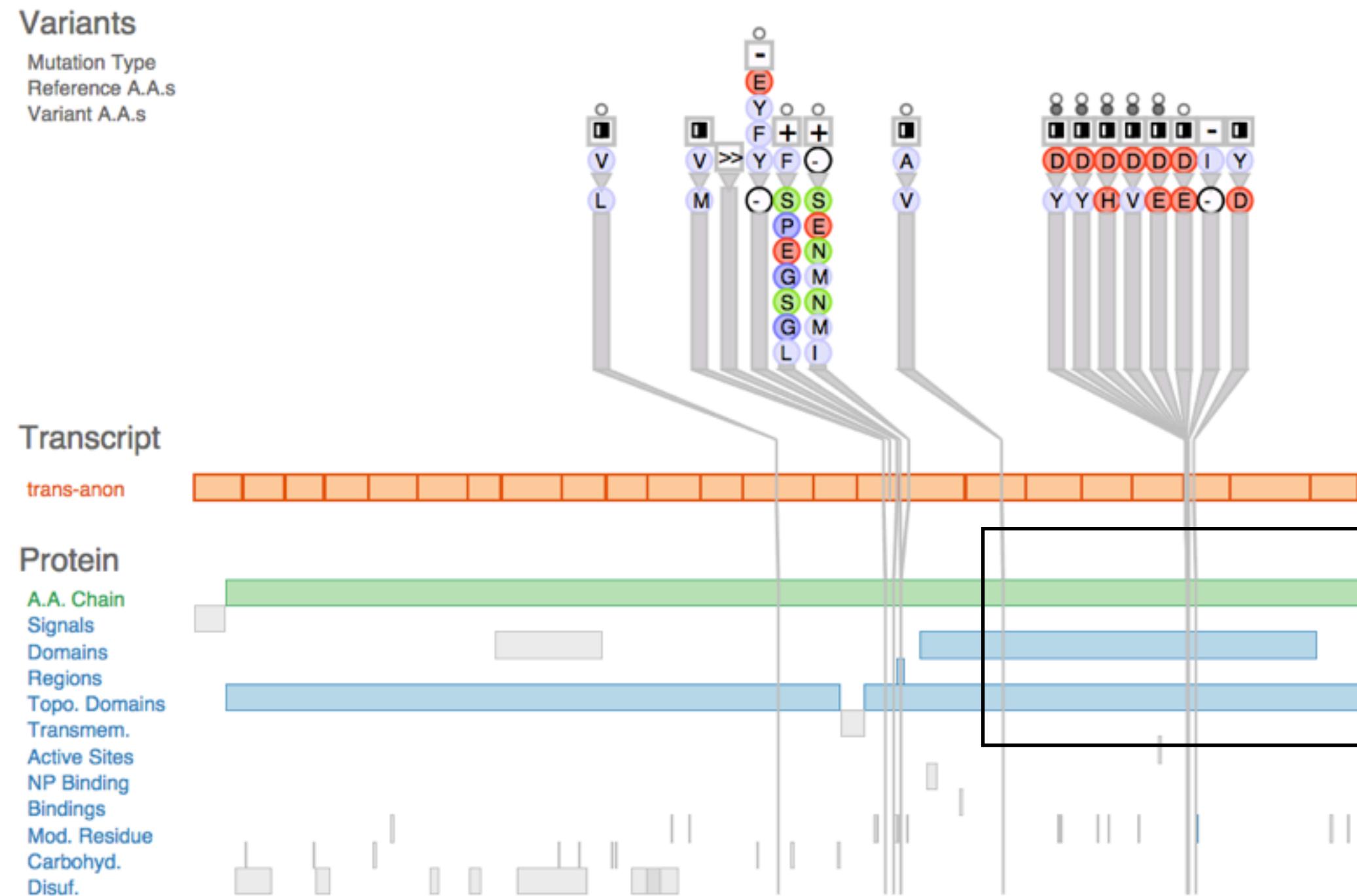
# Highly scored gene by sorting metric: known leukemia gene



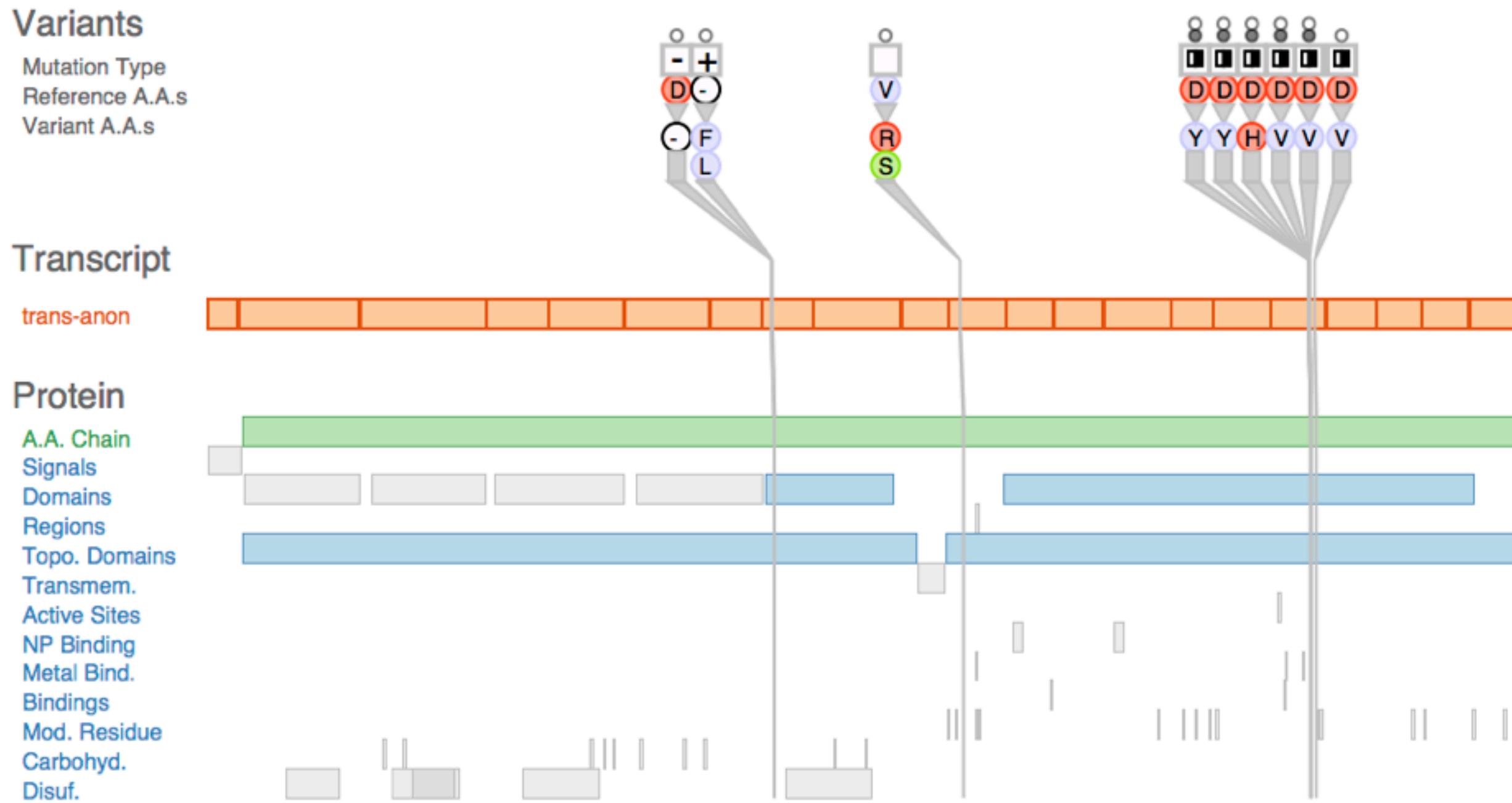
# Visual inspection reveals collocation of variants



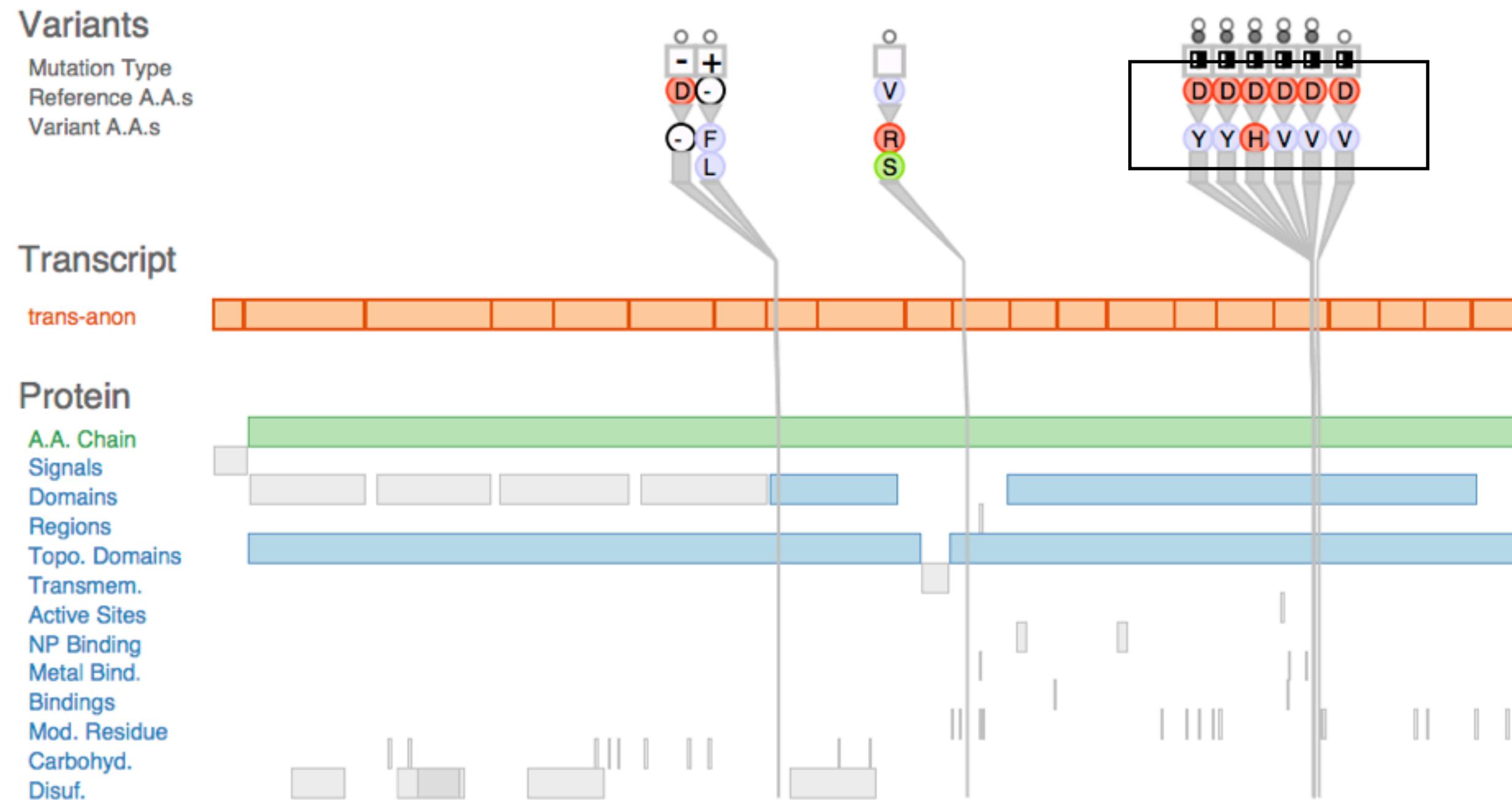
# Several functional protein regions affected



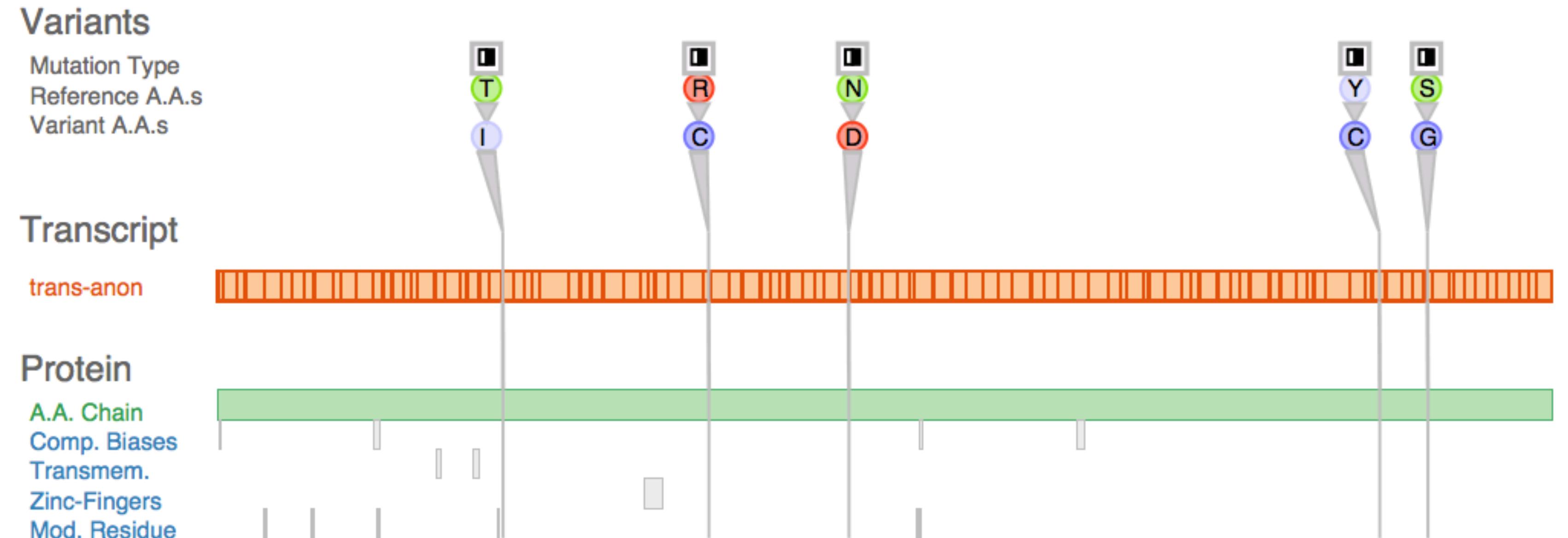
# Highly scored by metric: not previously known, good candidate



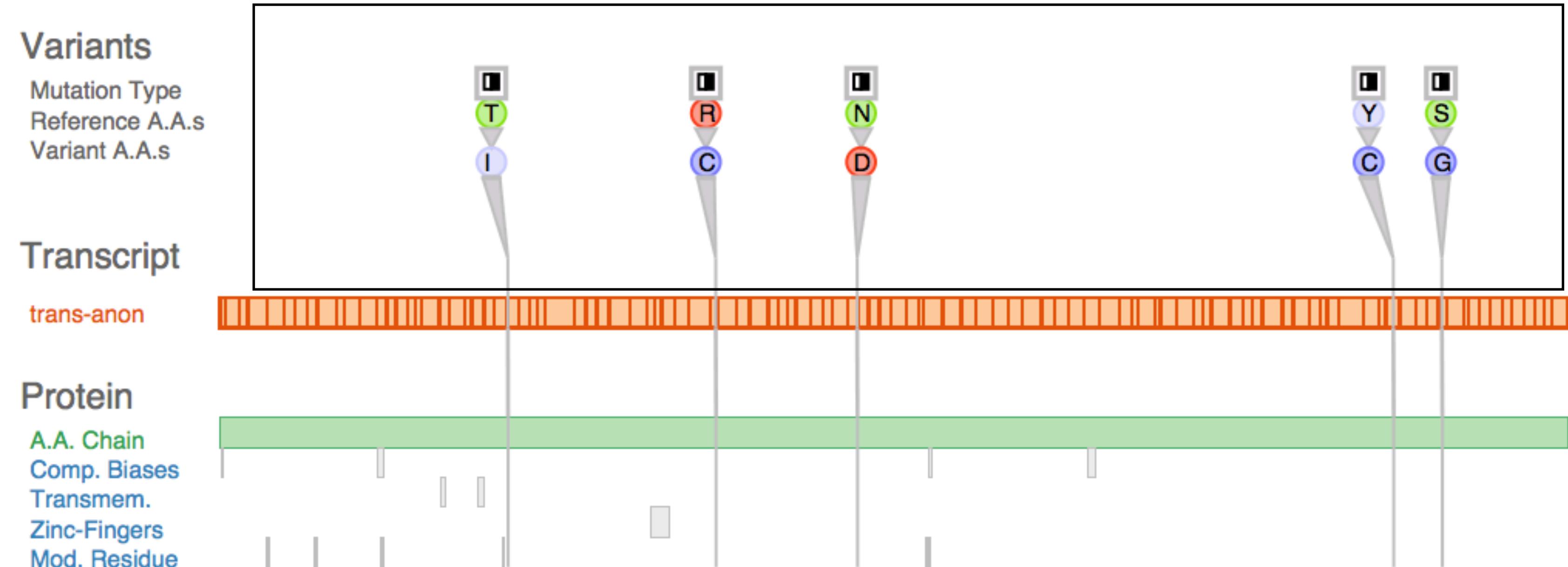
# Protein chemical class change evident



# In contrast, low scoring gene



# No collocation of variants



# Mostly unaffected protein regions

## Variants

Mutation Type  
Reference A.A.s  
Variant A.A.s



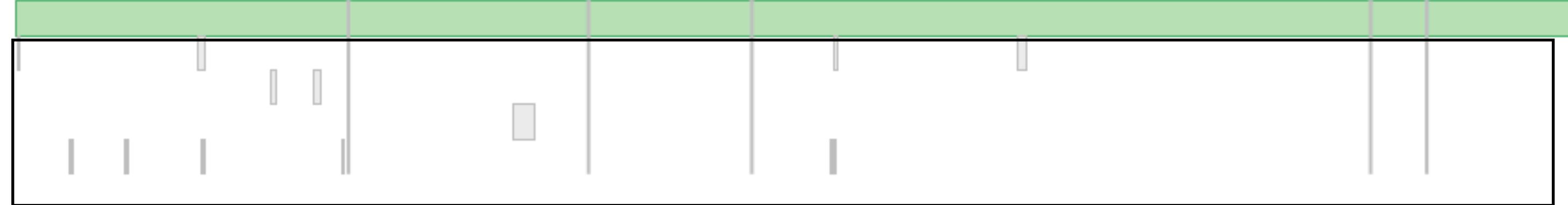
## Transcript

trans-anon



## Protein

A.A. Chain  
Comp. Biases  
Transmem.  
Zinc-Fingers  
Mod. Residue



# Methods

# Phase I: Winnow and Cast

5 months



- embedded within GSC for all stages
- **winnow stage**
  - considered and ruled out many potential collaborators
- **cast stage**
  - gatekeeper (PI)
  - two front-line analysts (postdocs)



more at:

Design Study Methodology: Reflections from the Trenches and from the Stacks.  
Sedlmair, Meyer, Munzner. *IEEE TVCG* 18(12): 2431-2440, 2012 (Proc. InfoVis 2012).

# Phase 2: Core Design

5 months



- main task abstraction
  - discover gene
- semi-structured interviews
  - every week for 1 hr
- iterative refinement
  - 8 data sketches deployed



# Phase 3: Two More Tasks

1 month



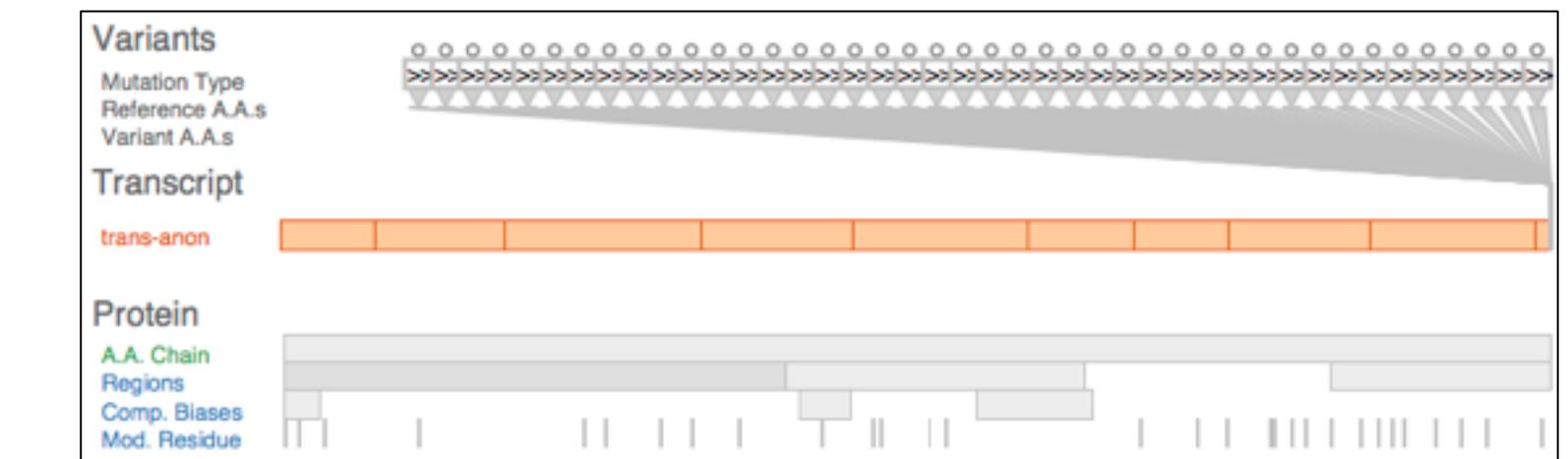
- two new analysts
  - connected by enthusiastic gatekeeper



- new task abstractions

- compare patients
- debug pipeline

- transferrable with minimal changes



## Phase 4: Reflect and write

3 months



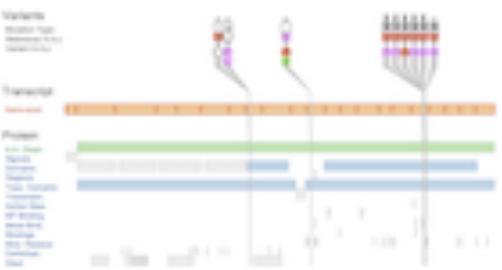
- abstraction innovation
  - data abstraction: highly filtered *transcript coordinates* (vs genome coordinates)
- guidelines
  - specialize first, generalize later
    - good for domains with complex data
  - high-level considerations
    - identifying scales of interest
    - what to visually encode directly vs what to support through interaction
    - when (and how) to eliminate navigation

# Themes, Revisited

- what and why to show: task and data abstraction
  - task and data commonalities cross-cut domains
- how to show: visual encoding and interaction idioms
  - RelEx: reduce memory load with interaction
  - VariantView: reduce interaction load with better visual encoding
- transferability from design studies
  - DSM: reflection to confirm/refute/refine/propose guidelines

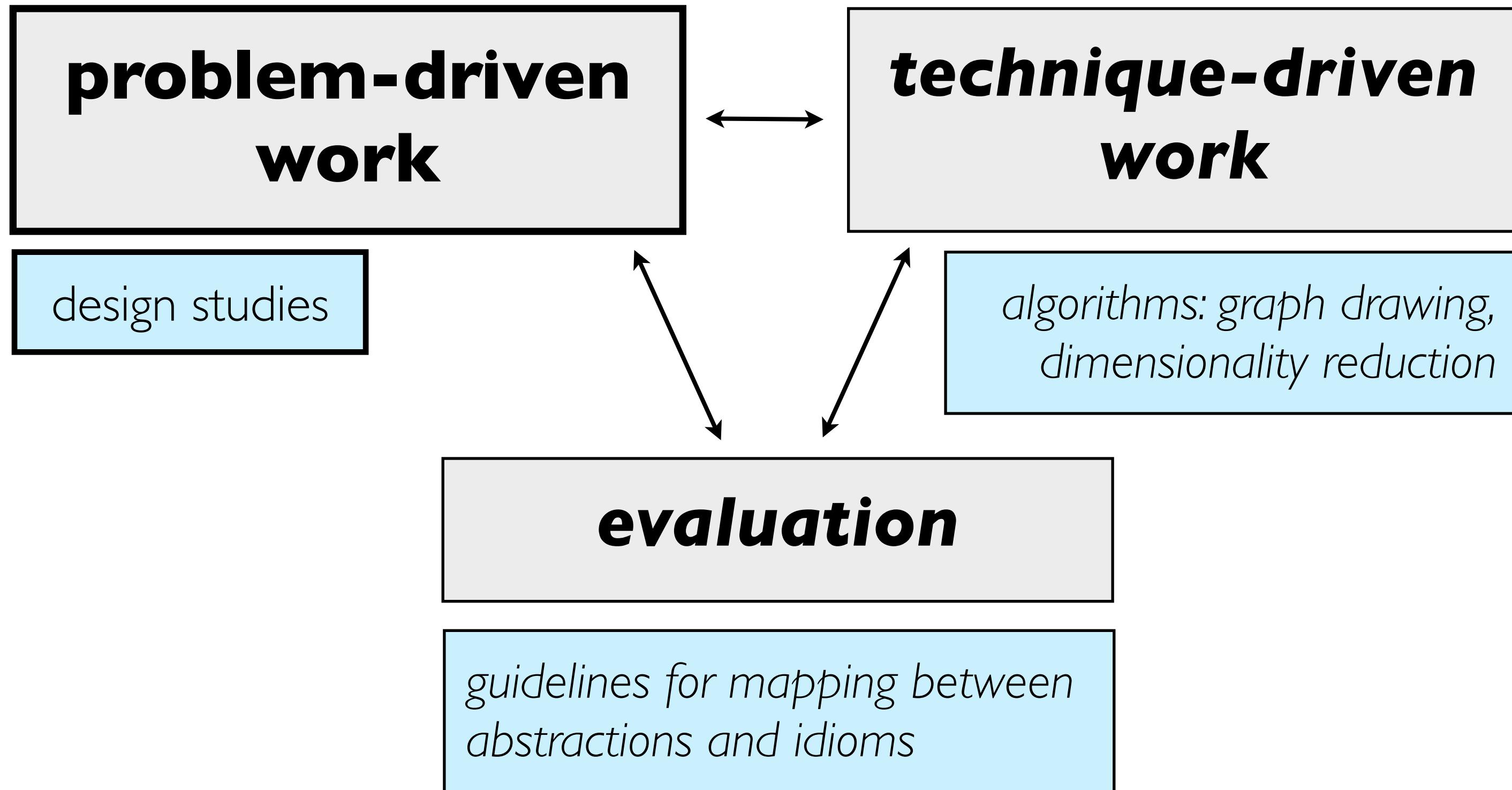


RelEx  
in-car networks



VariantView  
genomics

# A Different Trio: Research Interests



# Further Information

- further info
  - <http://www.cs.ubc.ca/~tmm/talks.html#chicago14> (this talk, and many others)
  - <http://www.cs.ubc.ca/group/infovis> (papers, software, videos)
  - <http://www.cs.ubc.ca/~tmm/courses/infovis> (course: readings, lectures)
  - <http://www.cs.ubc.ca/~tmm/courses/infovis/book> (book: to appear)
- open source software downloads
  - <http://www.cs.ubc.ca/labs/imager/tr/2013/VariantView/VariantViewSoftware/>
- acknowledgements
  - funding: NSERC, NSF
  - joint work: all co-authors
    - Andreas Butz, Annika Frank, Joel Ferstay, Miriah Meyer, Cydney Nielsen, Michael Sedlmair
  - feedback on this talk
    - Matthew Brehmer, Stephen Ingram